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

(19) World Intellectual Property Organization
International Bureau

(43) International Publication Date
3 January 2013 (03.01.2013)

WIPO | PCT

(10) International Publication Number
WO 2013/000100 A1

(51) International Patent Classification:
C12Q 1/68 (2006.01)

(21) International Application Number:
PCT/CN2011/001070

(22) International Filing Date:
29 June 2011 (29.06.2011)

(25) Filing Language: English

(26) Publication Language: English

(71) Applicant *(for all designated States except US)*: **BGI SHENZHEN CO., LIMITED** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN).

(72) Inventors; and

(75) Inventors/Applicants *(for US only)*: **JIANG, Fuman** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN). **CHEN, Huifei** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN). **CHAI, Xianghua** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN). **YUAN, Yuying** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN). **ZHANG, Xiuqing** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN). **CHEN, Fang** [CN/CN]; Main Building 11F-3, Beishan Industrial Zone, Beishan Road 146, Yantian District, Shenzhen, Guangdong 518083 (CN).

(74) Agent: **PEKSUNG INTELLECTUAL PROPERTY LTD.**; 908 Shining Tower, 35 Xueyuan Road, Haidian District, Beijing 100191 (CN).

(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, 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, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, 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, ML, MR, NE, SN, TD, TG).

Published:

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

(54) Title: NONINVASIVE DETECTION OF FETAL GENETIC ABNORMALITY

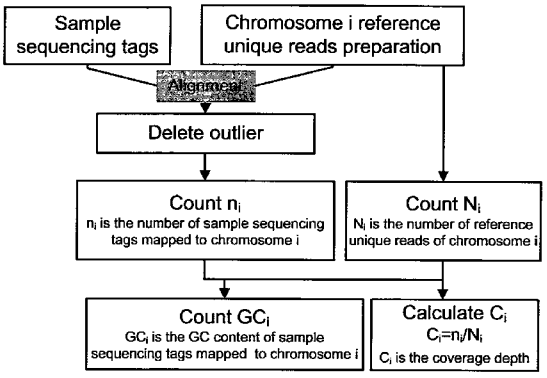

Figure 1

(57) Abstract: The current invention is directed to methods for noninvasive detection of fetal genetic abnormalities by large-scale sequencing of nucleotides from maternal biological sample. Further provided are methods to remove GC bias from the sequencing results according to the difference in GC content of a chromosome. The current invention not only makes the detection much more accurate but also represents a comprehensive method for fetal aneuploidy detection including sex chromosome disorders such as XO, XXX,XXY, and XYY, etc.

WO 2013/000100 A1