

Title (en)
METHOD OF PRENATAL DIAGNOSIS

Title (de)
VERFAHREN ZUR PRÄNATALEN DIAGNOSE

Title (fr)
PROCÉDÉ DE DIAGNOSTIC PRÉNATAL

Publication
EP 3149202 A1 20170405 (EN)

Application
EP 15736606 A 20150526

Priority
• IT TO20140414 A 20140526
• IB 2015053926 W 20150526

Abstract (en)
[origin: WO2015181718A1] Method of prenatal diagnosis comprising the steps of: acquiring (1) reads of fragments of genomic libraries obtained from circulating free total DNA in a sample of maternal plasma, the DNA representing a plurality of chromosomes; aligning (2) the reads with respect to a predetermined reference genome obtaining a first dataset (SAM); converting (4) the first dataset (SAM) into a second dataset (BAM) so as to allow sorting thereof according to chromosomal coordinates and successive duplicate removal, obtaining a third dataset containing all the reads aligned and devoid of duplicates; subdividing (6) at least one chromosome into windows of predetermined dimensions, so as to count the reads which have a "mapping validity" greater than or equal to a threshold value in each reading window, obtaining a total number of above-threshold reads (NW); normalizing, for each reading window, the total number of above-threshold reads (NW) with respect to the total number (Ntot) of reads of the fragment of genomic library obtaining a first normalized datum (NW,lib); normalizing (8), for each reading window, the first normalized datum (NW,lib) with respect to a systematic error due to the content of G and C bases, obtaining a second normalized datum (NW,lib,GC); calculating (10), for each chromosome, a median of all the second normalized data (NW,lib,GC) obtaining a final value (Nji); calculating (12) an aneuploidy parameter (Z) for at least one chromosome; determining the aneuploidy parameter (Z) of at least one chromosome so as to evaluate whether the sample of maternal plasma is aneuploid.

IPC 8 full level
C12Q 1/68 (2006.01); **G16B 20/10** (2019.01); **G16B 20/20** (2019.01); **G16B 30/10** (2019.01); **G16B 40/00** (2019.01)

CPC (source: EP US)
C12Q 1/6869 (2013.01 - EP); **G16B 20/00** (2019.01 - EP); **G16B 20/10** (2019.01 - EP US); **G16B 20/20** (2019.01 - EP US); **G16B 30/00** (2019.01 - EP); **G16B 30/10** (2019.01 - EP US); **C12Q 2537/159** (2013.01 - EP); **C12Q 2537/16** (2013.01 - EP); **C12Q 2537/165** (2013.01 - EP); **G16B 40/00** (2019.01 - EP US)

Citation (search report)
See references of WO 2015181718A1

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