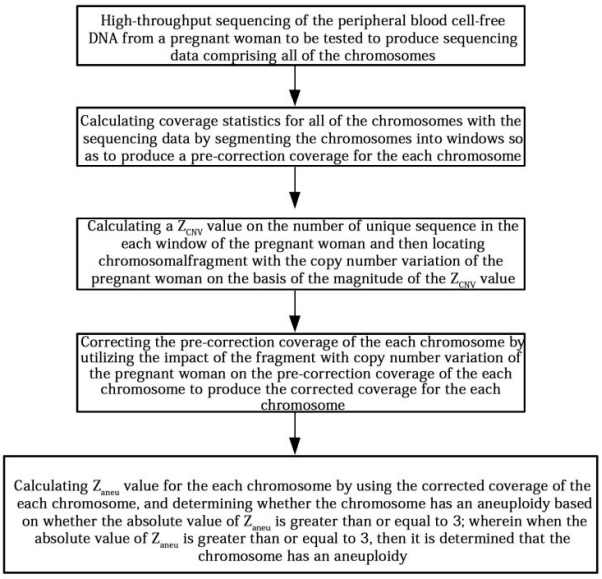

High-throughput sequencing of the peripheral blood cell-free DNA from a pregnant woman to be tested to produce sequencing data comprising all of the chromosomes
Calculating coverage statistics for all of the chromosomes with the sequencing data by segmenting the chromosomes into windows so as to produce a pre-correction coverage for the each chromosome
Calculating a Z_{CNV} value on the number of unique sequence in the each window of the pregnant woman and then locating chromosomalfragment with the copy number variation of the pregnant woman on the basis of the magnitude of the Z_{CNV} value
Correcting the pre-correction coverage of the each chromosome by utilizing the impact of the fragment with copy number variation of the pregnant woman on the pre-correction coverage of the each chromosome to produce the corrected coverage for the each chromosome
Calculating Z_{aneu} value for the each chromosome by using the corrected coverage of the each chromosome, and determining whether the chromosome has an aneuploidy based on whether the absolute value of Z_{aneu} is greater than or equal to 3; wherein when the absolute value of Z_{aneu} is greater than or equal to 3, then it is determined that the chromosome has an aneuploidy

fig.1

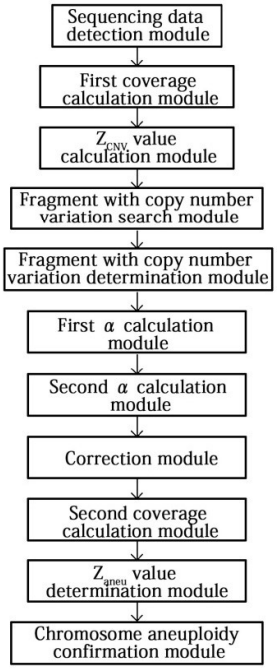

Sequencing data detection module
First coverage calculation module
Z_{cNV} value calculation module
Fragment with copy number variation search module
Fragment with copy number variation determination module
First α calculation module
Second α calculation module
Correction module
Second coverage calculation module
Z_{aneu} value determination module
Chromosome aneuploidy confirmation module

fig.2

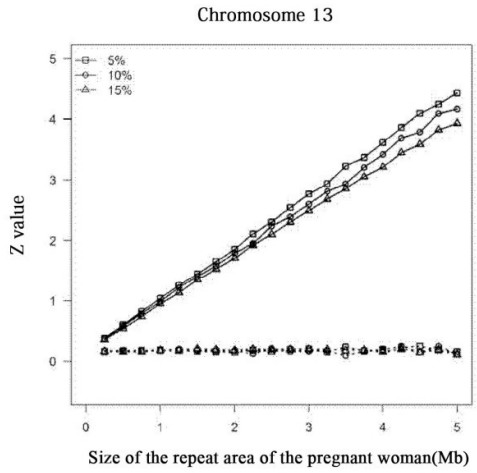

Chromosome 13
5%
10%
15%
Z value
0
1
2
3
4
5
Size of the repeat area of the pregnant woman(Mb)

fig.3A

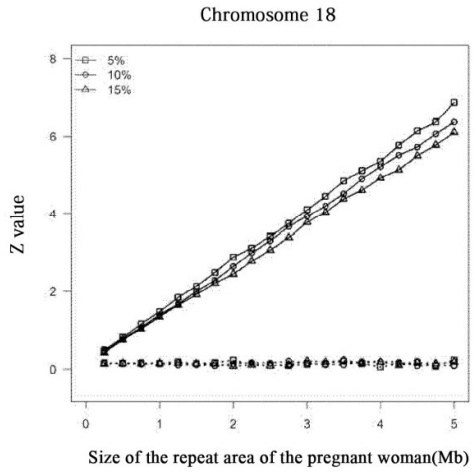

Chromosome 18
5%
10%
15%
Z value
0
2
4
6
8
Size of the repeat area of the pregnant woman(Mb)

fig.3B

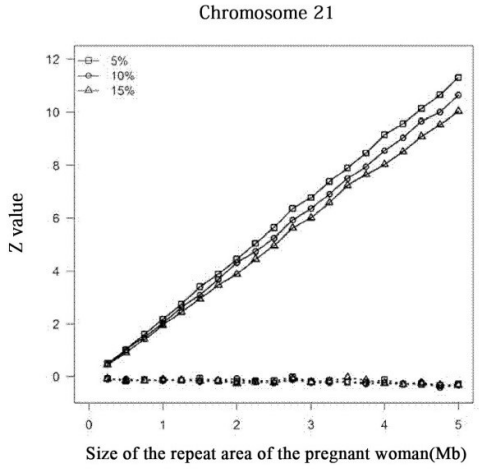

Chromosome 21
5%
10%
15%
Z value
Size of the repeat area of the pregnant woman(Mb)

fig.3C

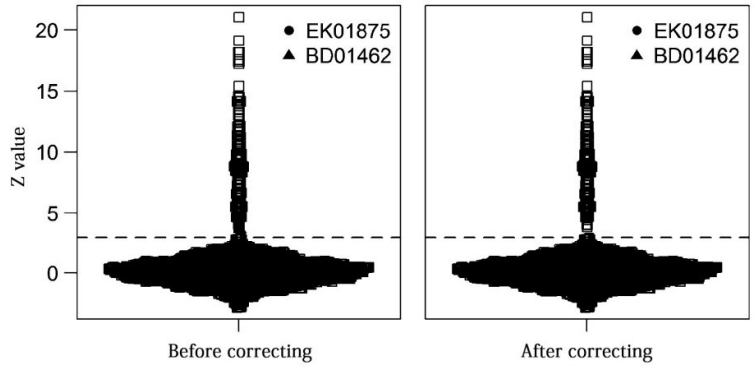

EK01875
BD01462
Z value
Before correcting
After correcting

fig.4

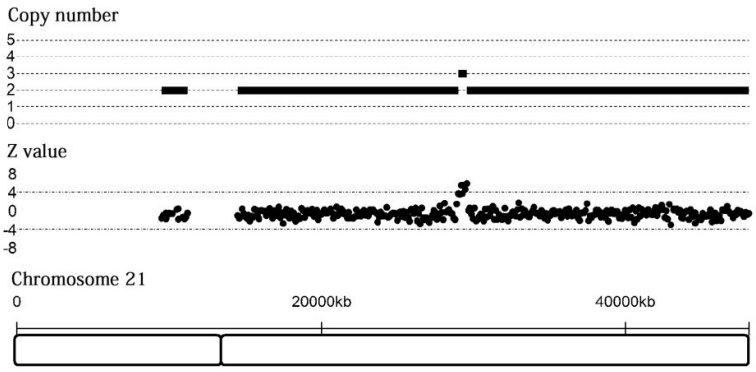

Copy number
5
4
3
2
1
0
Z value
8
4
0
-4
-8
Chromosome 21
0
20000kb
40000kb

fig.5

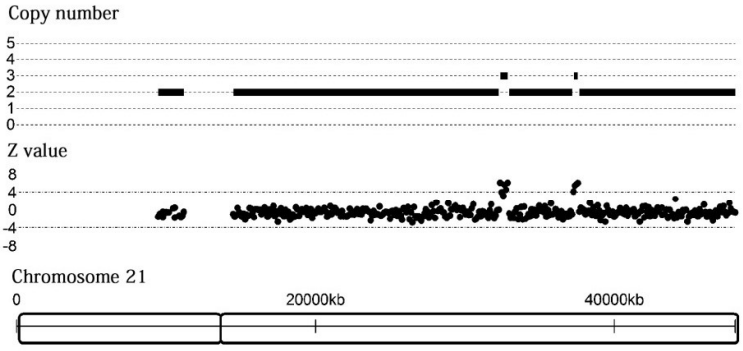

Copy number
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4
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2
1
0
Z value
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4
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-4
-8
Chromosome 21
0
20000kb
40000kb

fig.6