

Figure 1

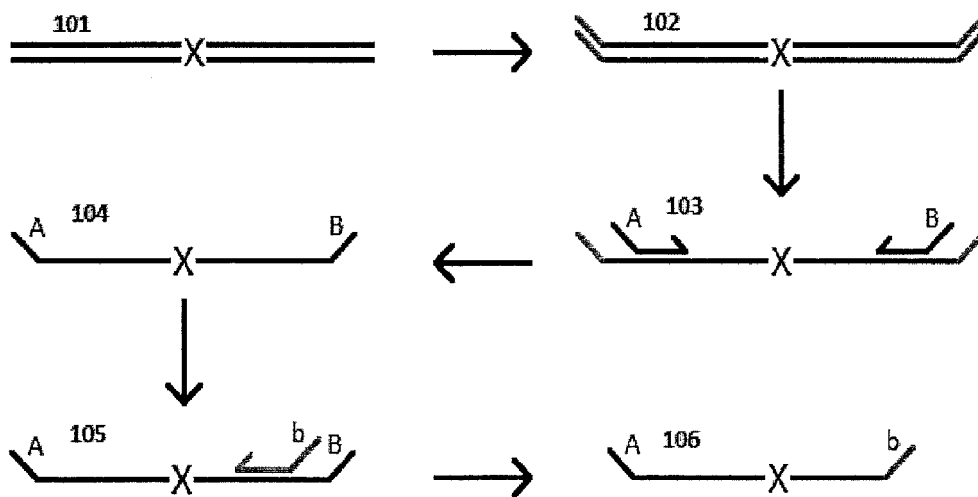


Figure 2

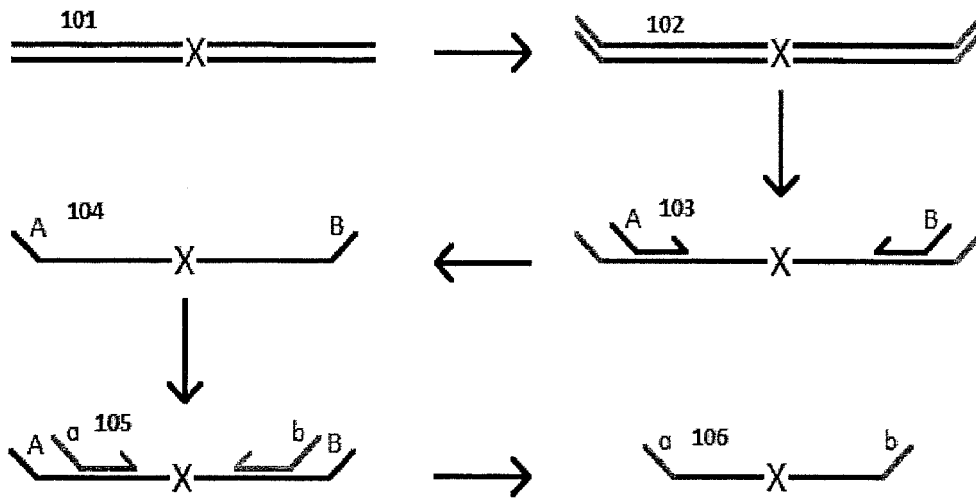


Figure 3

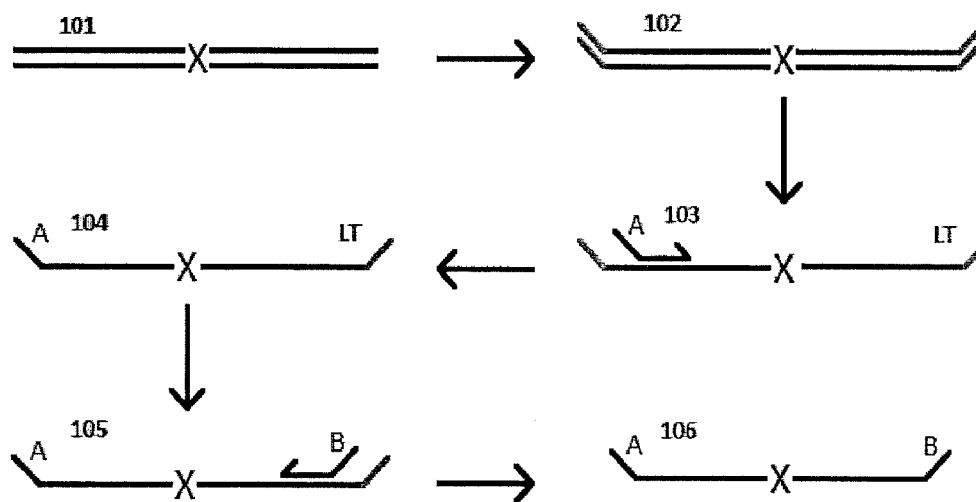


Figure 4

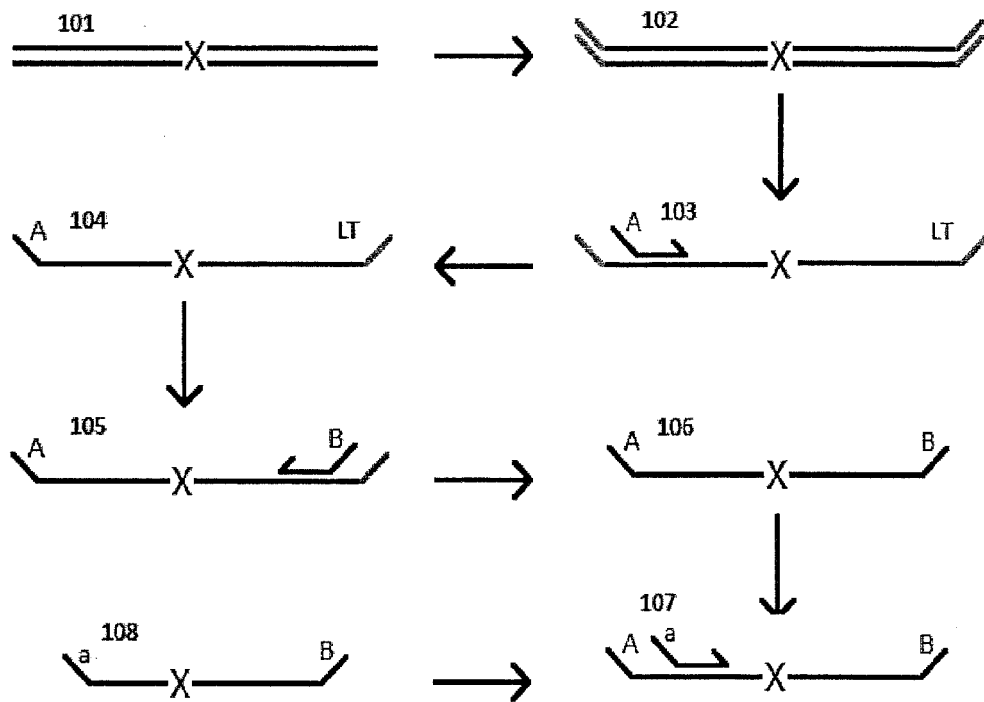


Figure 5

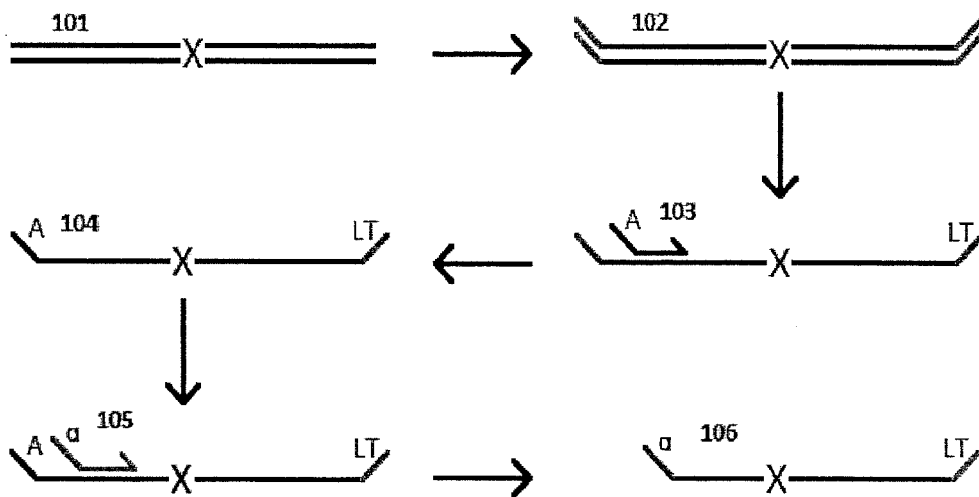


Figure 6

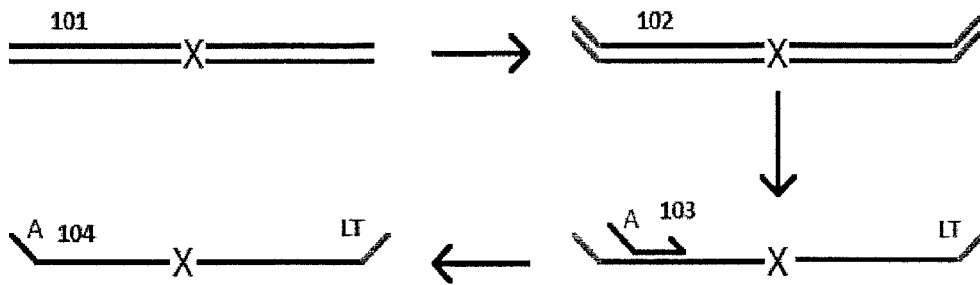


Figure 7

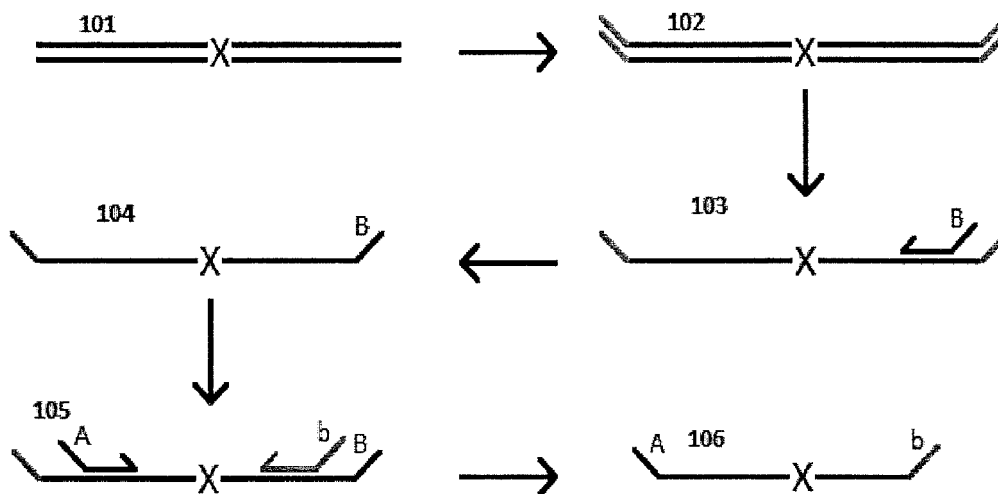


Figure 8

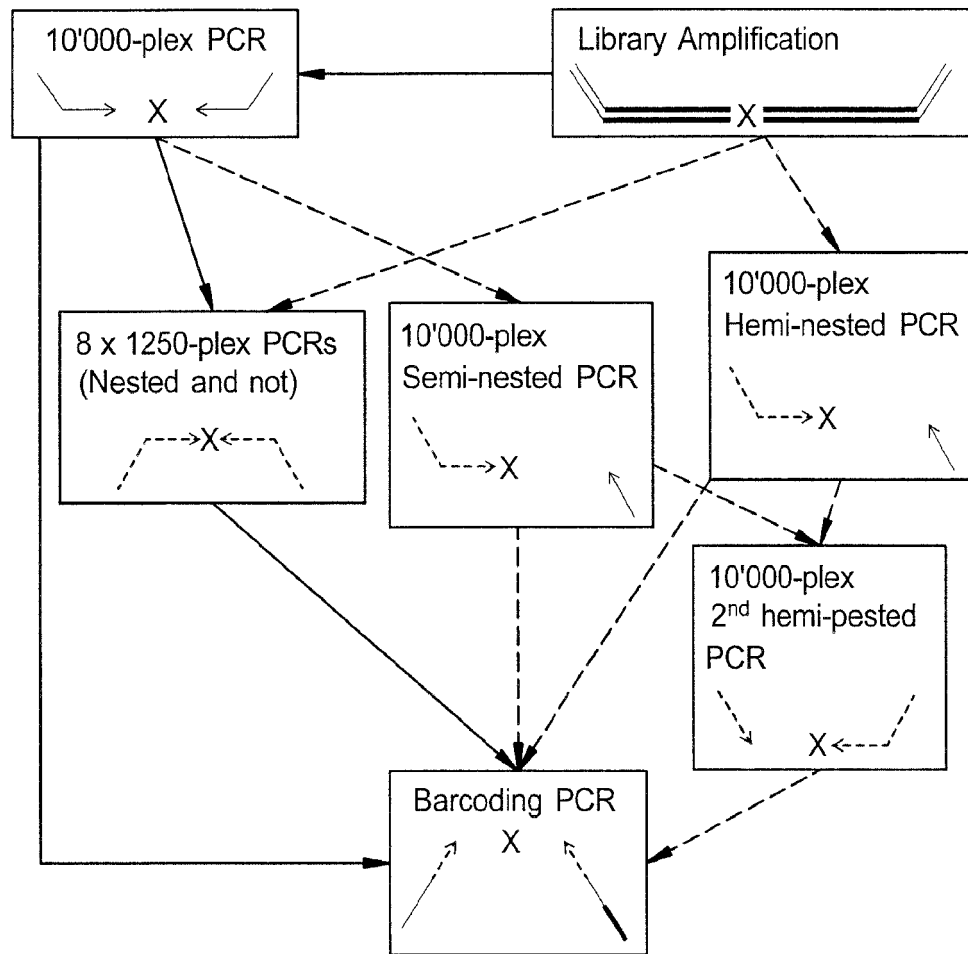


Figure 9

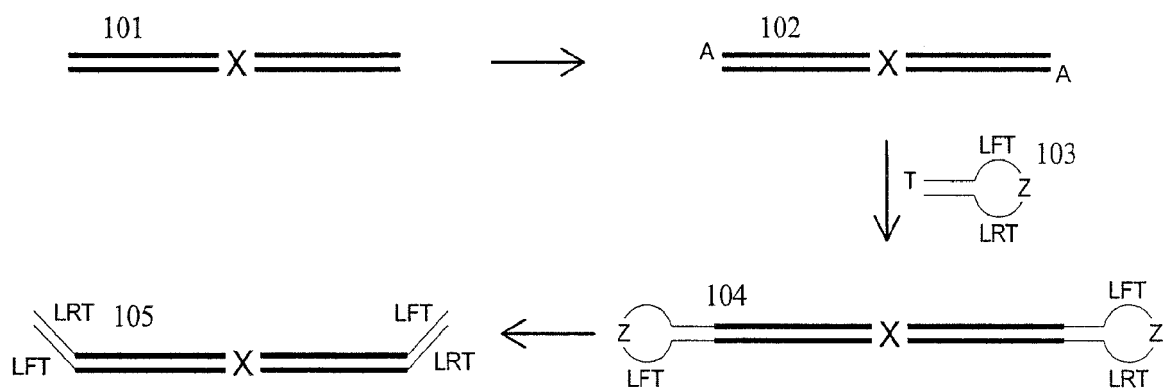
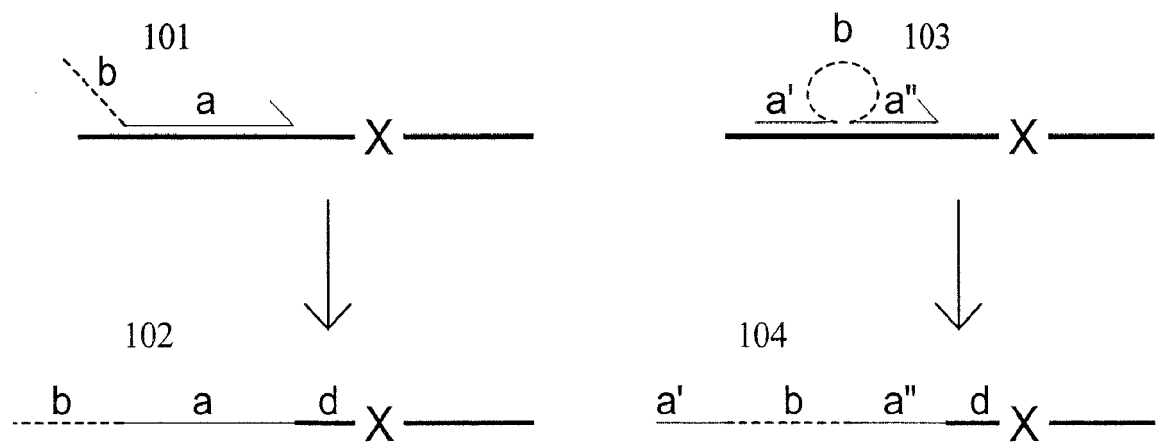
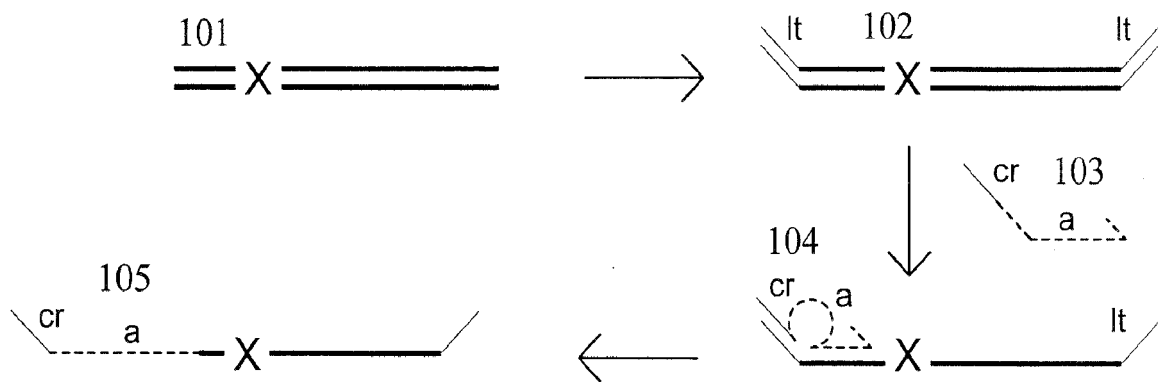


Figure 10

**Figure 11**

The sequencing-adaptor sequence is located inside the primer sequence and flanked by target specific sequence on both sides. 10 bases are target-specific at the 3'-end of each primer. Primers were tested successfully in real-time PCR. For sequencing this reduces the number of primer bases that need to be sequenced.			
rs8130564	int-tag 1.10	AACTCACATAGC <u>ACACGACGCTCTCCGATCT</u> TGCAAGCACA	seqid1
rs2832093	int-tag 2.10	TCCTCTGTG <u>ACACGACGCTCTCCGATCT</u> CCCTGCTCTT	seqid2
rs12011281	int-tag 3.10	tcctctctcACACGACGCTCTCCGATCTcGGGCTGTCA	seqid3
rs6719561	int-tag 4.10	TACATCCTTGAGACACGACGCTCTCCGATCTGCTGTGCAGT	seqid4
rs10187018	int-tag 5.10	tttgcttgagctACACGACGCTCTCCGATCTcgggagtttc	seqid5
rs10460481	int-tag 6.10	gtcttatggtggACACGACGCTCTCCGATCTcaaagccagt	seqid6
The sequencing-adaptor sequence is located inside the primer sequence and flanked by target specific sequence on both sides. The internal tag is formed into a hairpin structure by 10 complementary bases on either end. This brings the target-specific ends of the primer into close proximity and hinders unspecific binding to the "internal tag". 10 bases are target-specific at the 3'-end of each primer. Primers were tested successfully in real-time PCR.			
rs8130564	loop-int-tag 1.10	AACTCACATAGC <u>tgatcgggACACGACGCTCTCCGATCT</u> TGCAAGCACA	seqid7
rs2832093	loop-int-tag 2.10	TCCTCTGTG <u>tgatcgggACACGACGCTCTCCGATCT</u> CCCTGCTCTT	seqid8
rs12011281	loop-int-tag 3.10	tcctctcttgatcgggACACGACGCTCTCCGATCTcGGGCTGTCA	seqid9
rs6719561	loop-int-tag 4.10	TACATCCTTGAGtgatcgggACACGACGCTCTCCGATCTGCTGTGCAGT	seqid10
rs10187018	loop-int-tag 5.10	tttgcttgagcttgatcgggACACGACGCTCTCCGATCTcgggagtttc	seqid11
rs10460481	loop-int-tag 6.10	gtcttatggtggtgatcgggACACGACGCTCTCCGATCTcaaagccagt	seqid12

Figure 12

**Figure 13**

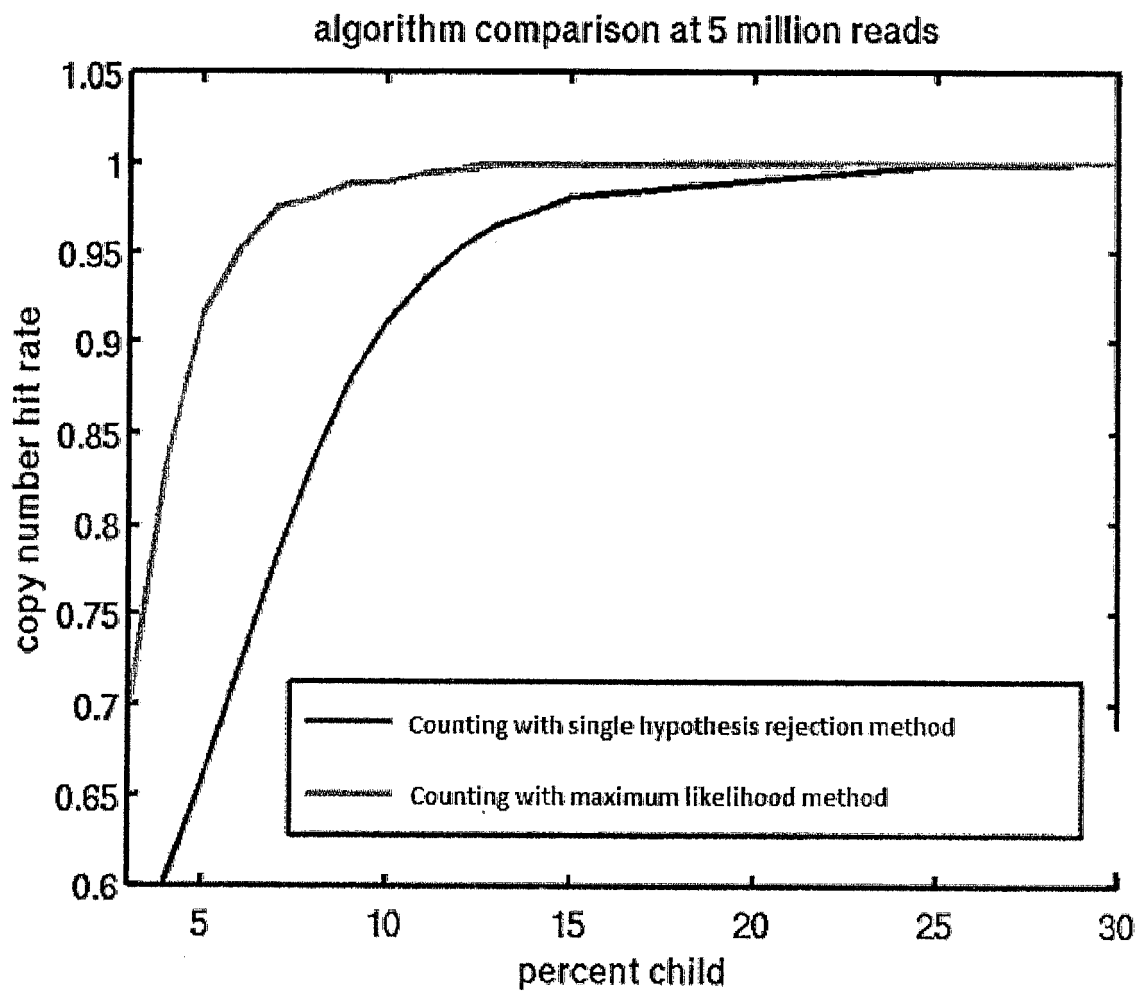


Figure 14

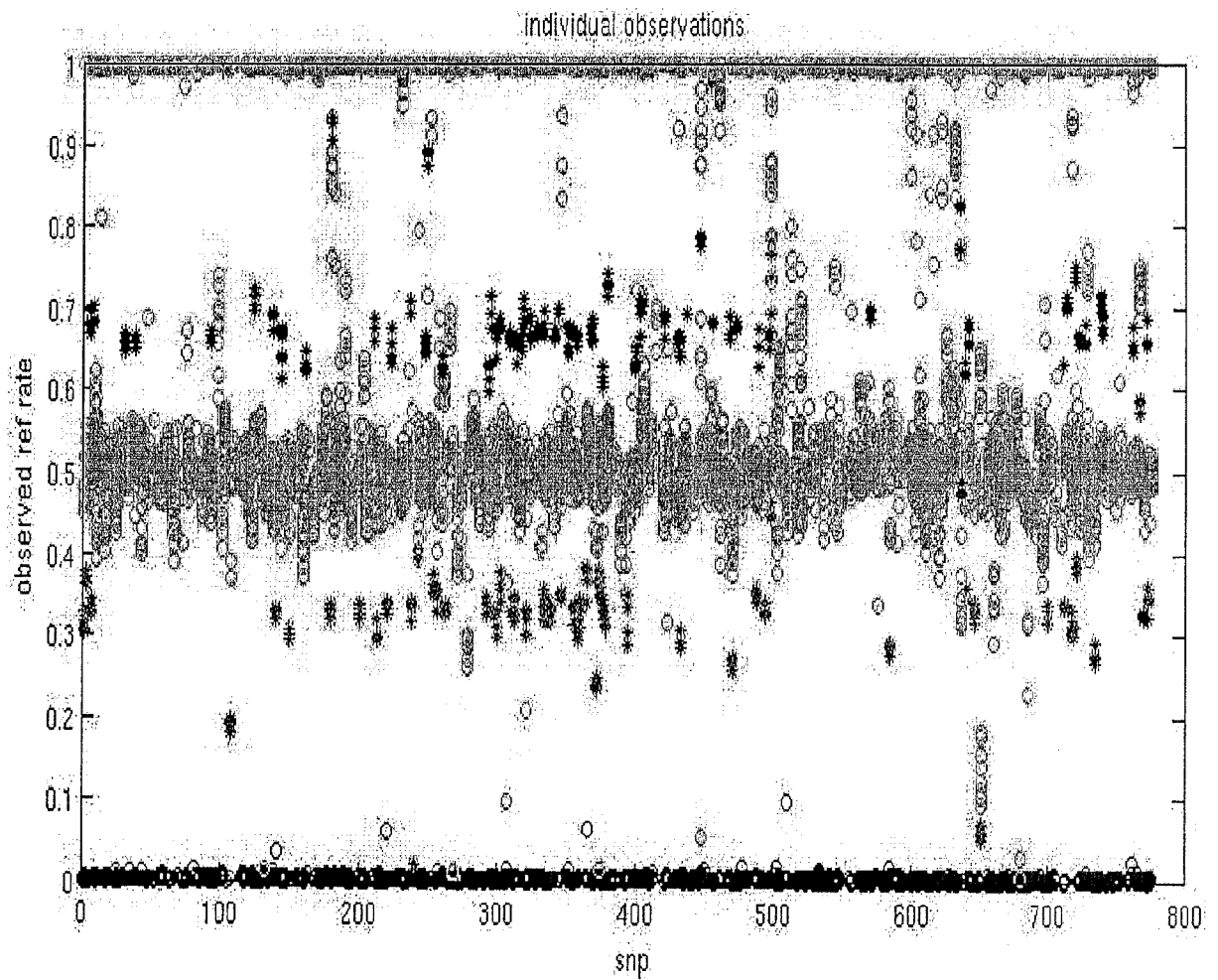


Figure 15

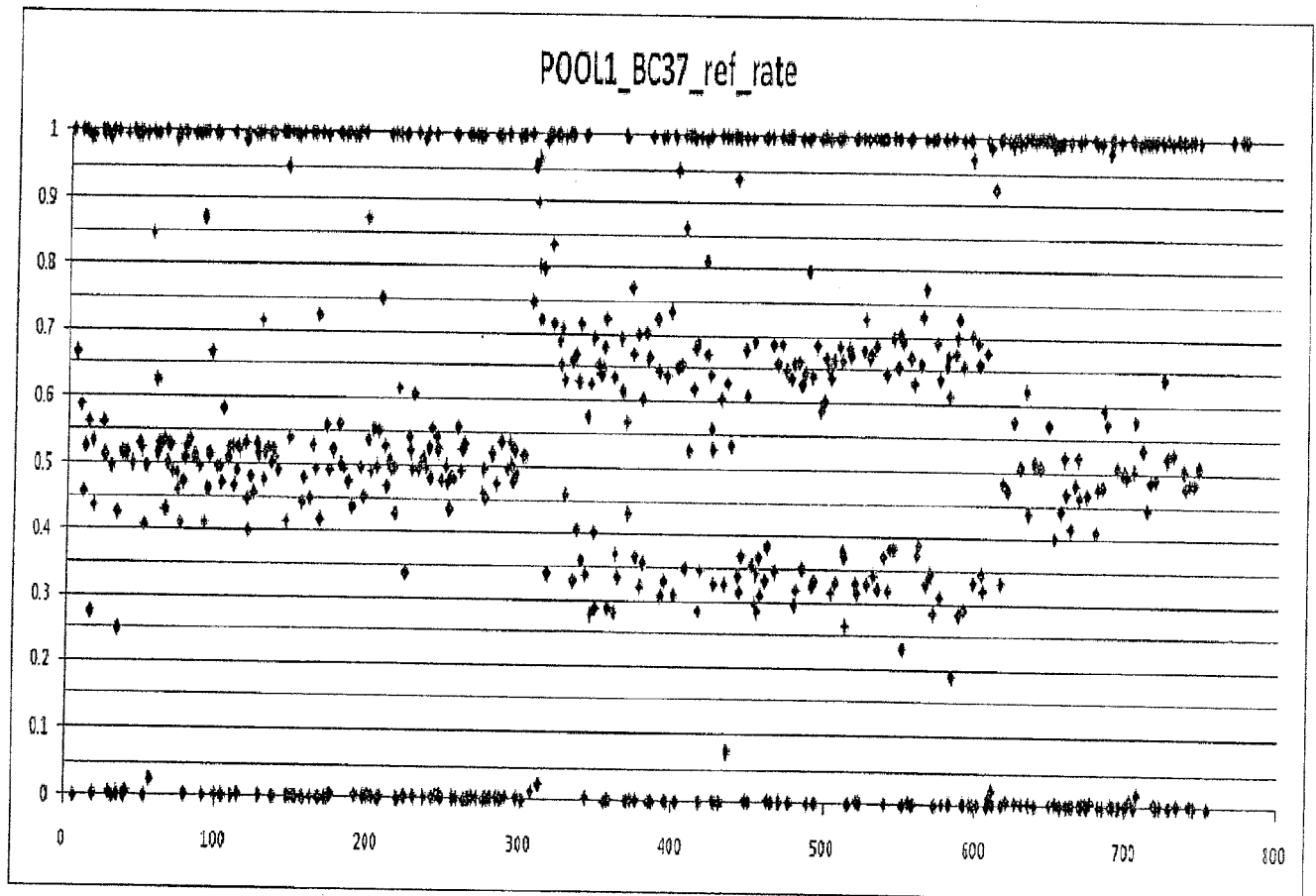


Figure 16

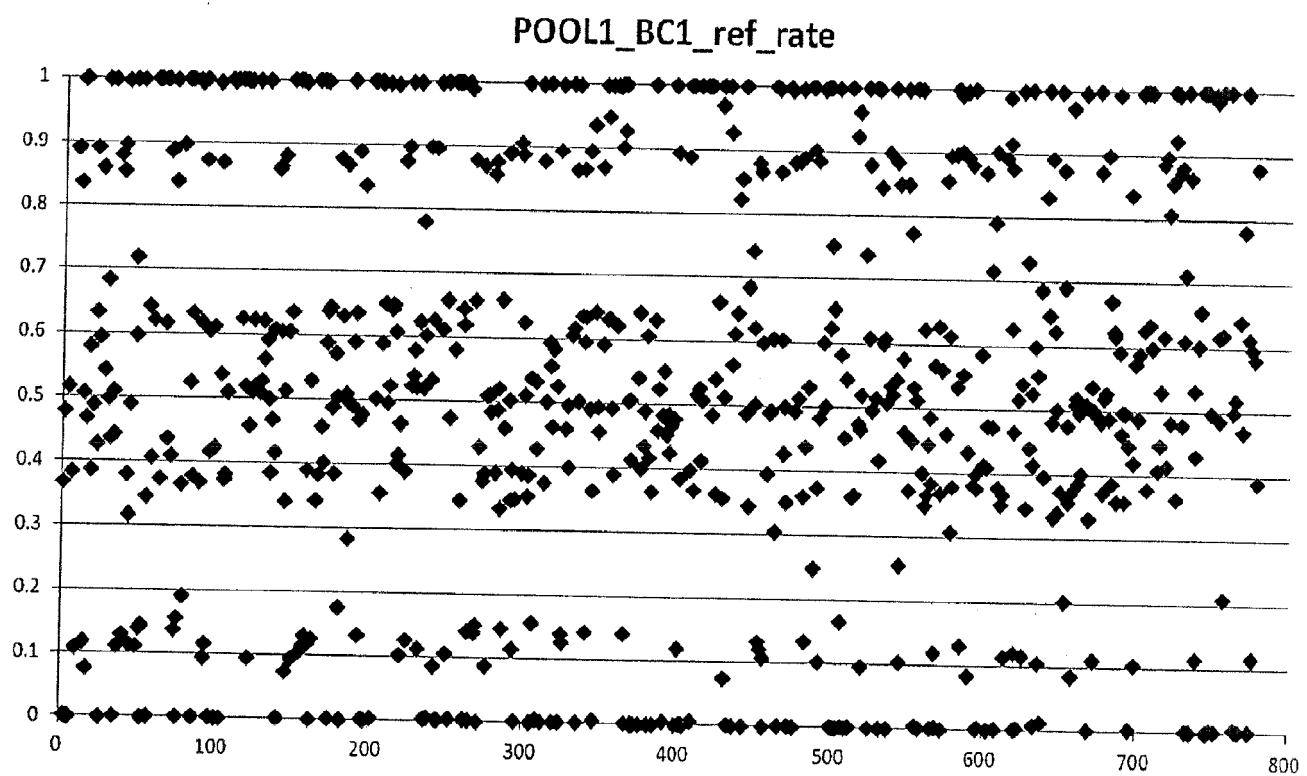


Figure 17A

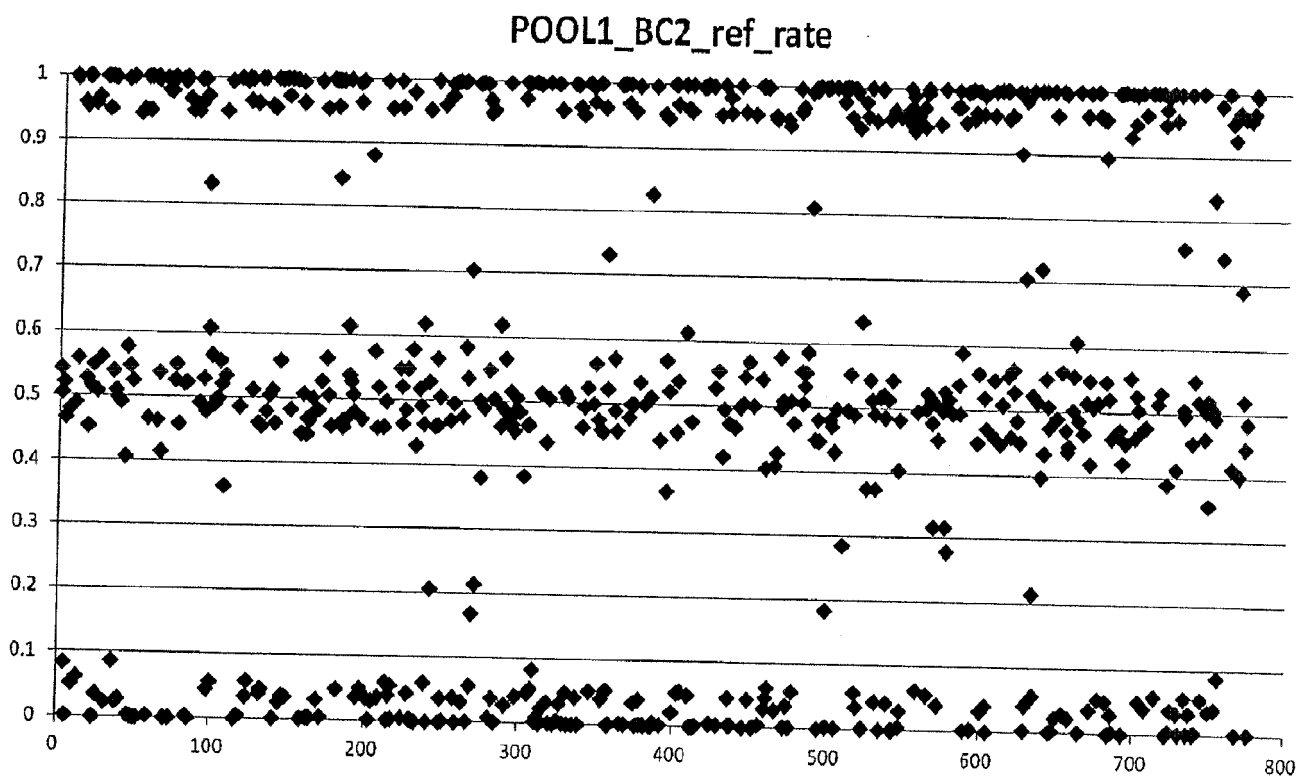


Figure 17B

No fetal DNA

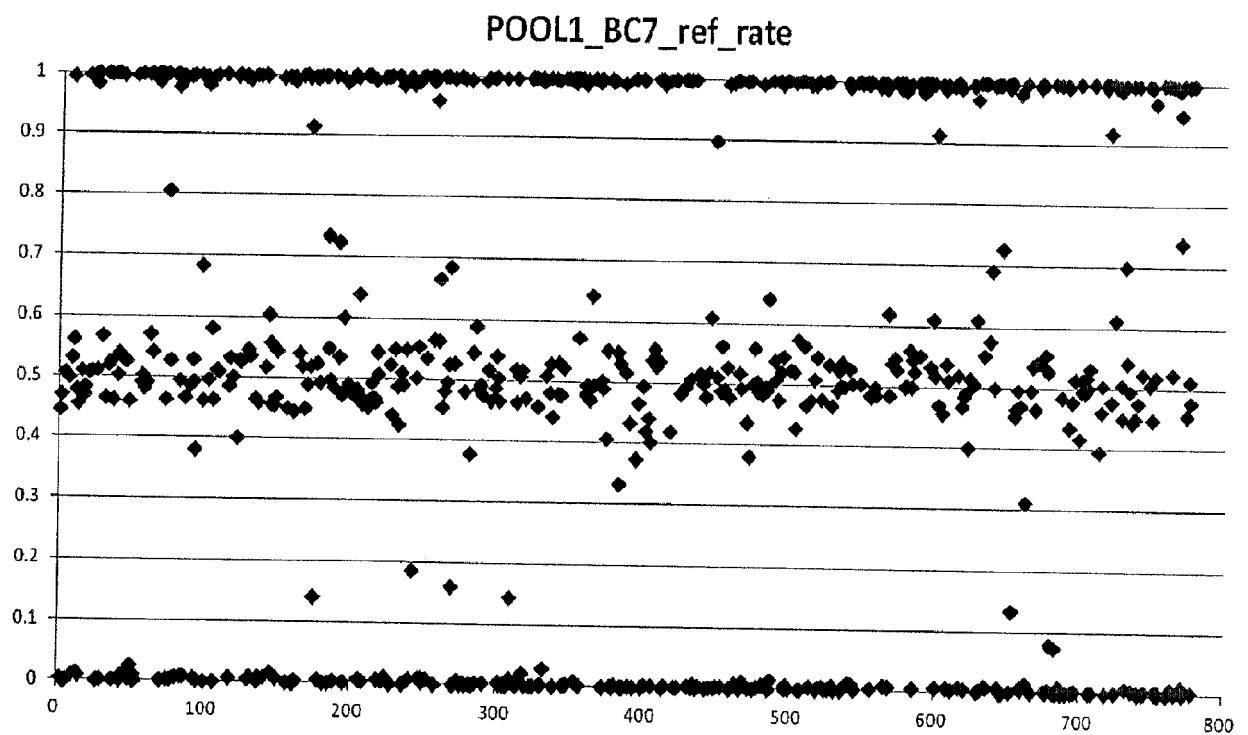


Figure 17C

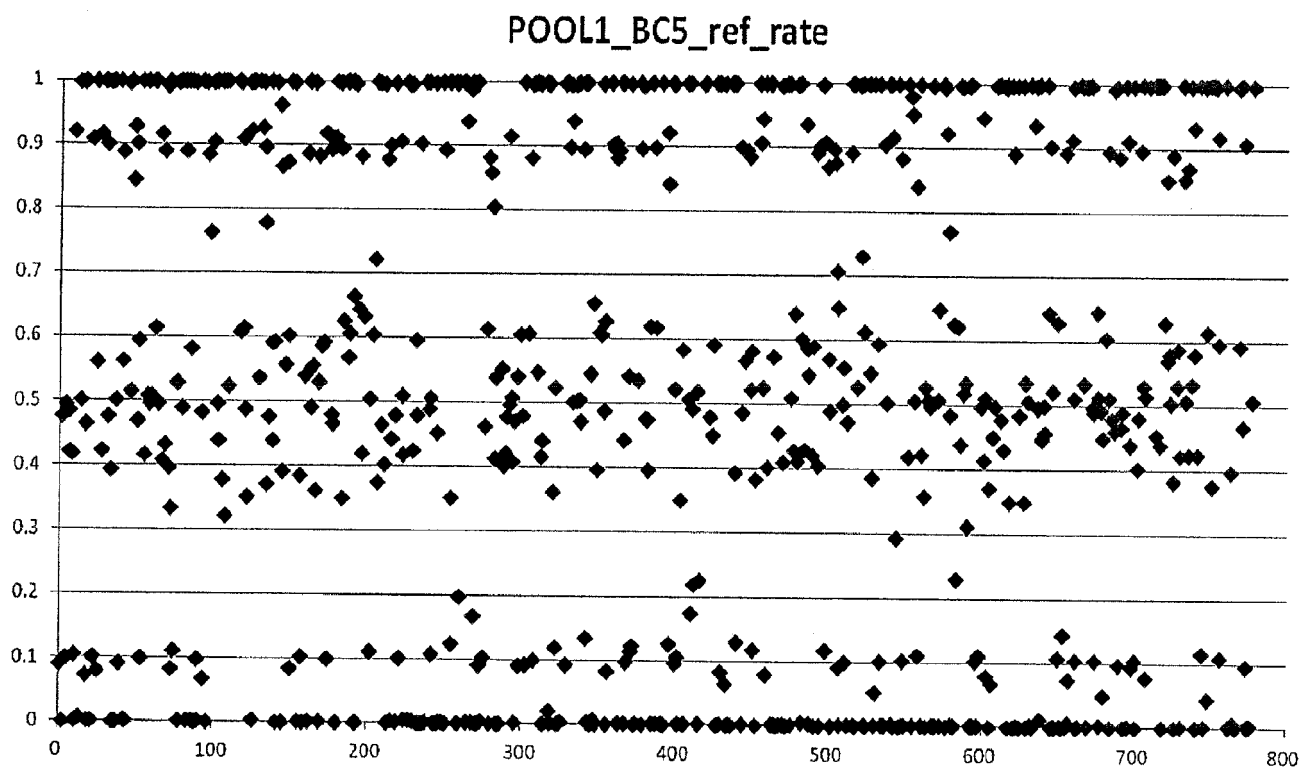
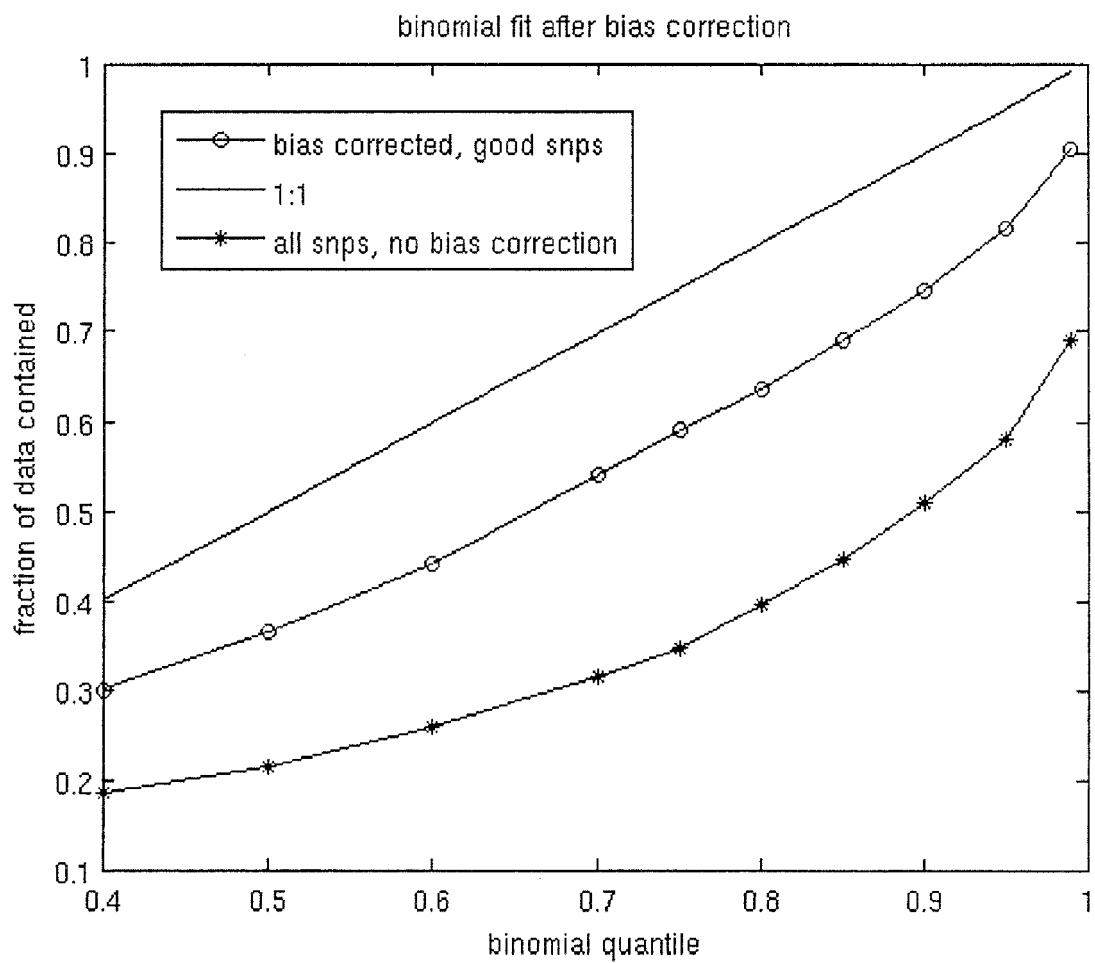


Figure 17D

**Figure 18**

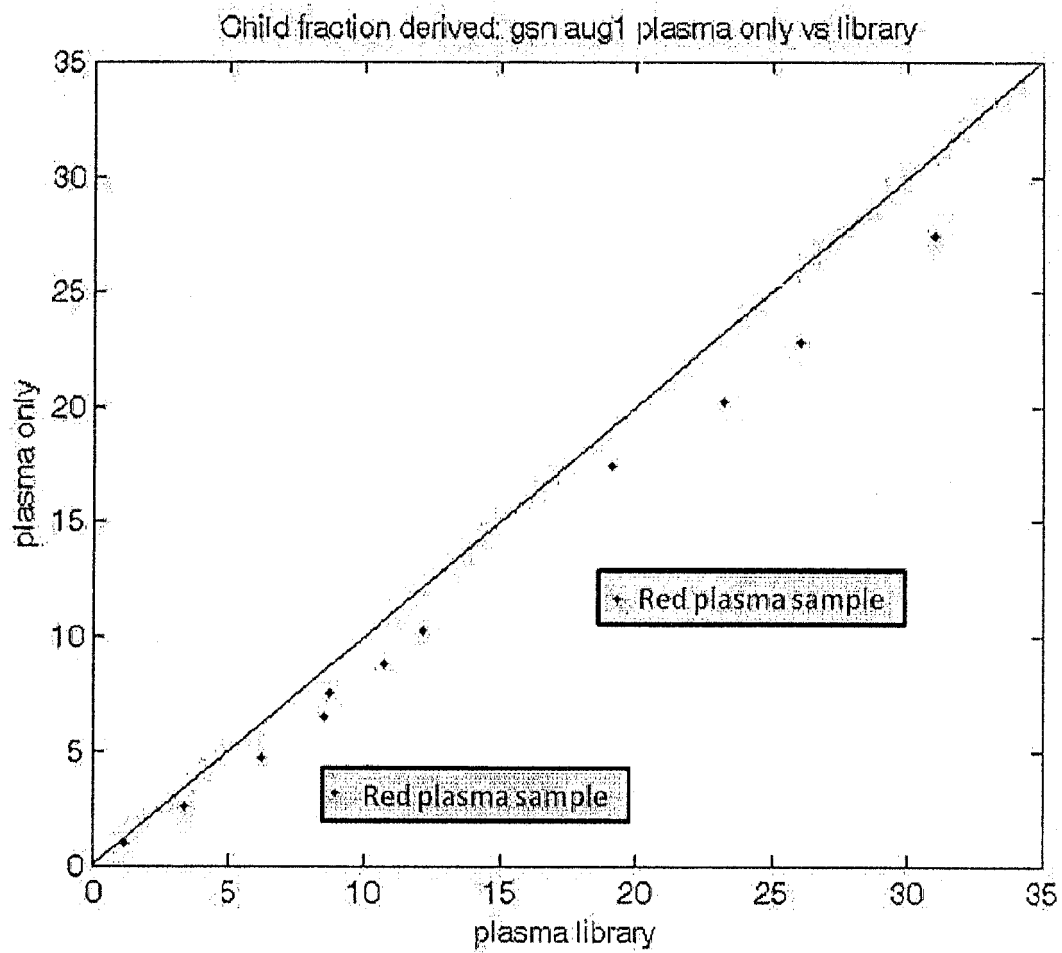


Figure 19

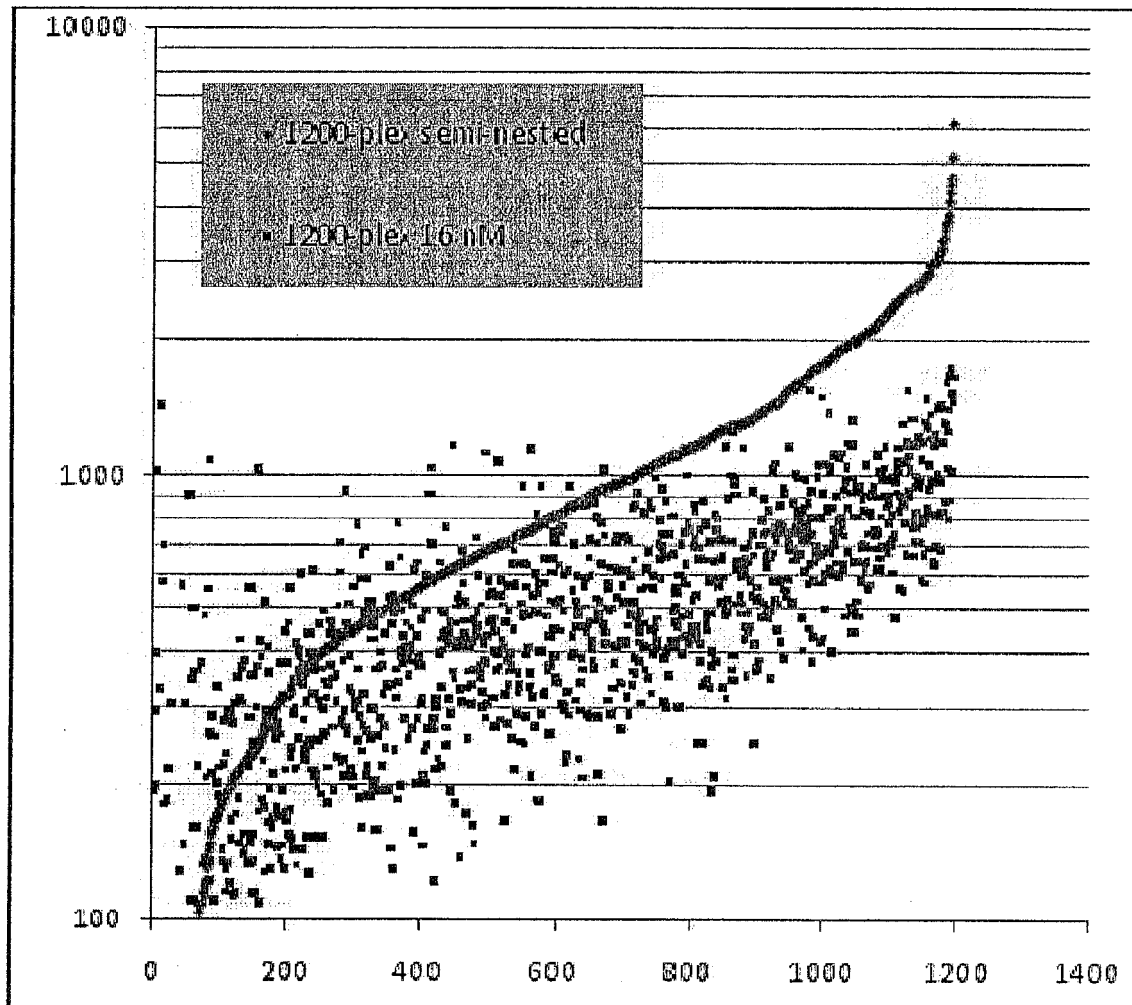


Figure 20

DOR range direct 1200-plex, 10 nM, 10 min TA

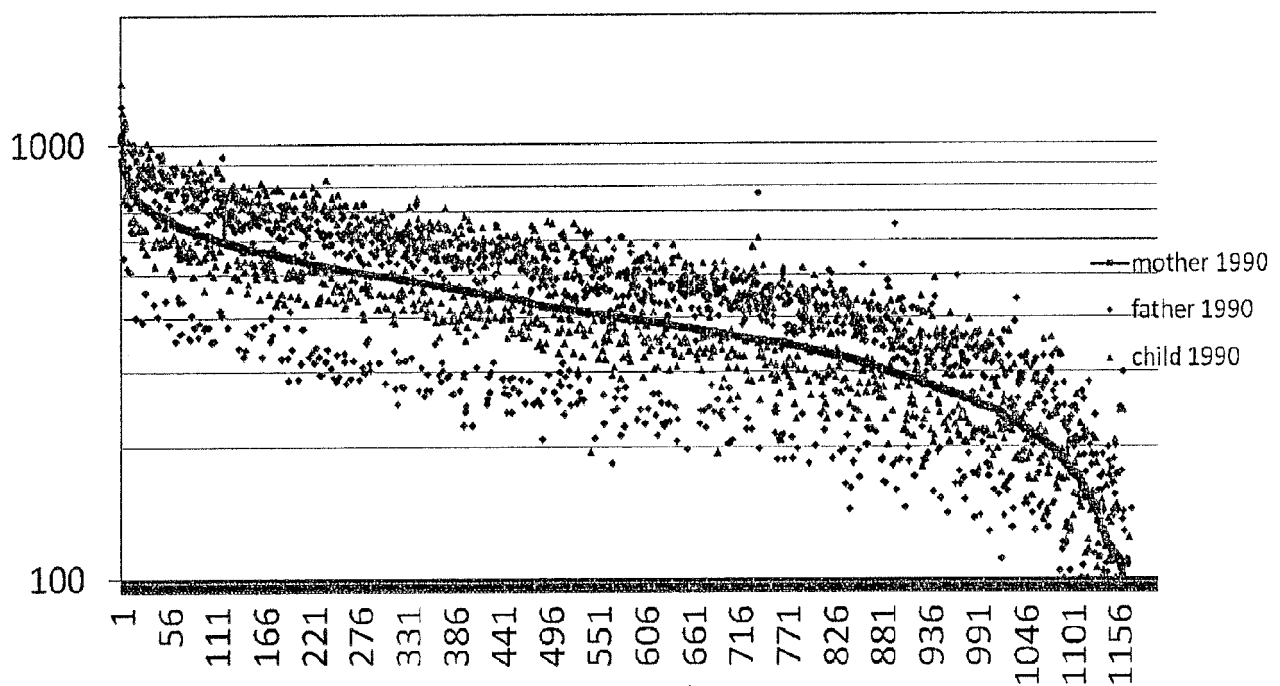
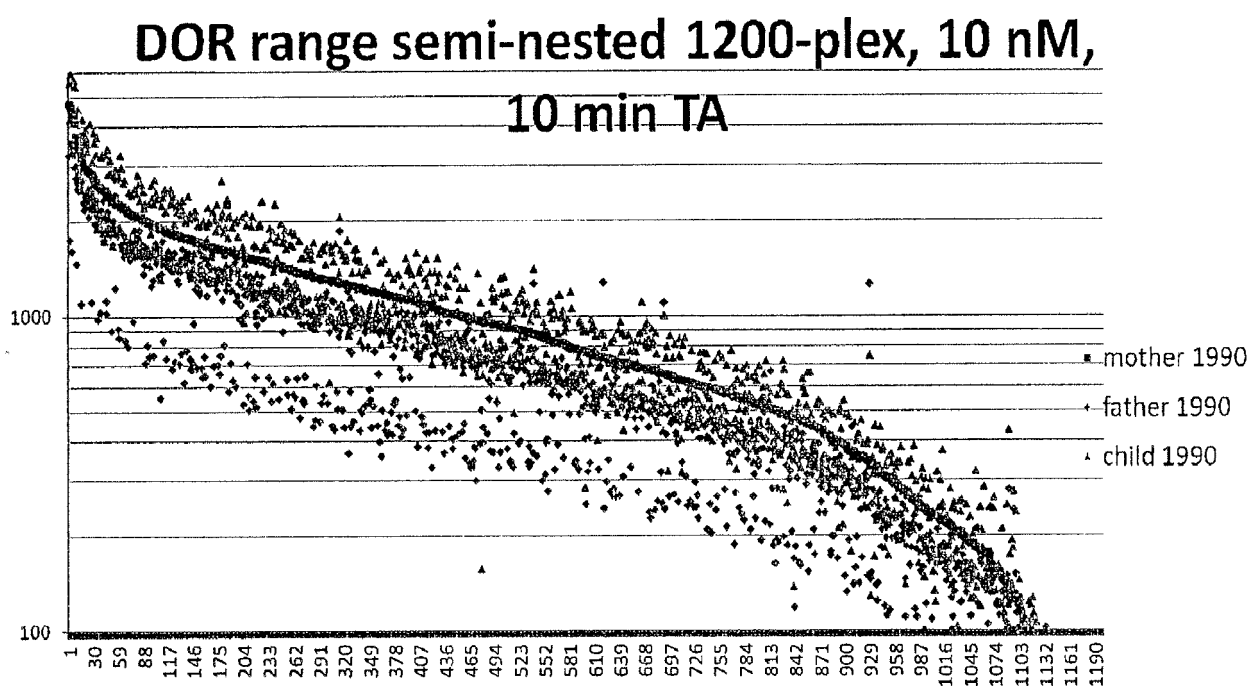


Figure 21

**Figure 22**

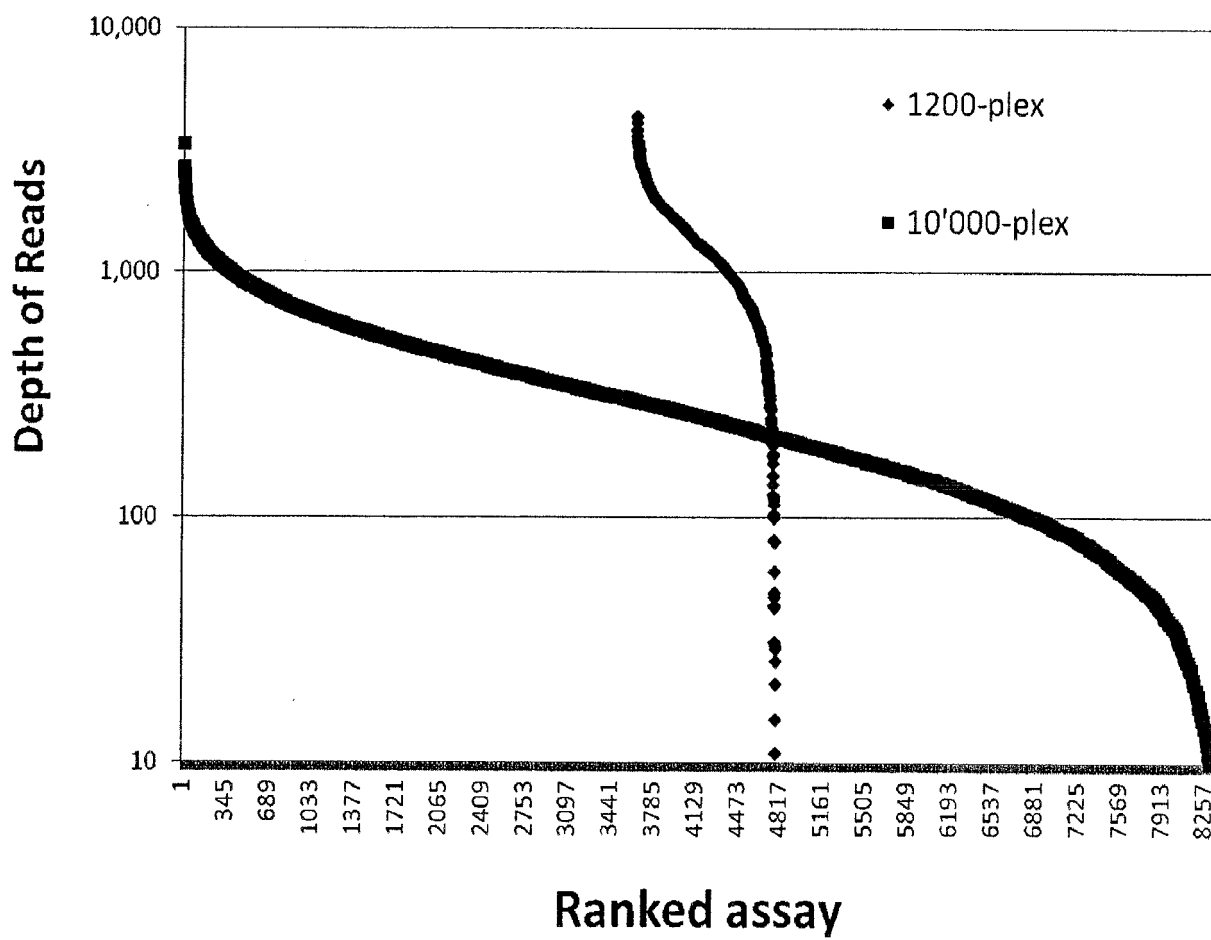


Figure 23

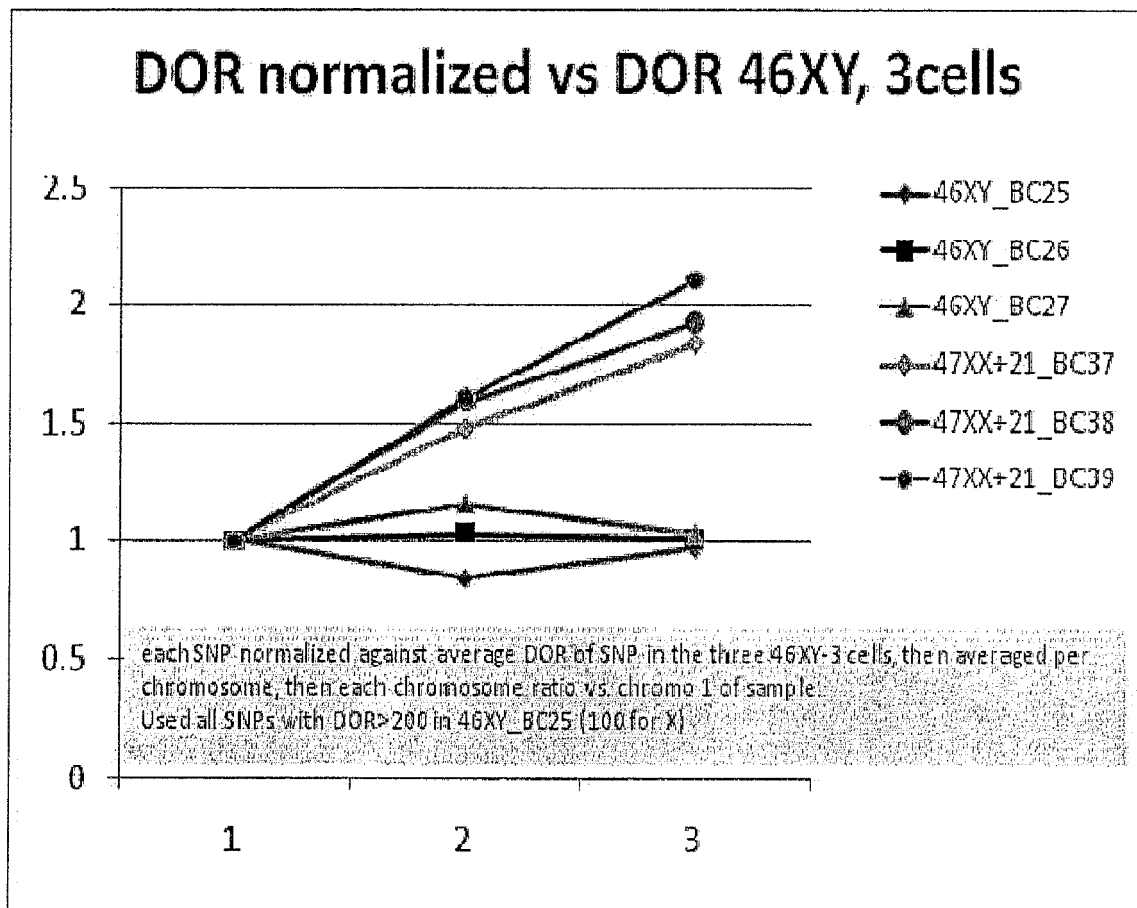


Figure 24

- Data from 3 cells with Arcturus: mini-PCR can detect trisomy 21 from 3 cells.
- Left: chromosome 1, middle chromosome 21 and right chromosome X.
- Sorted from left to right by DOR for each chromosome.

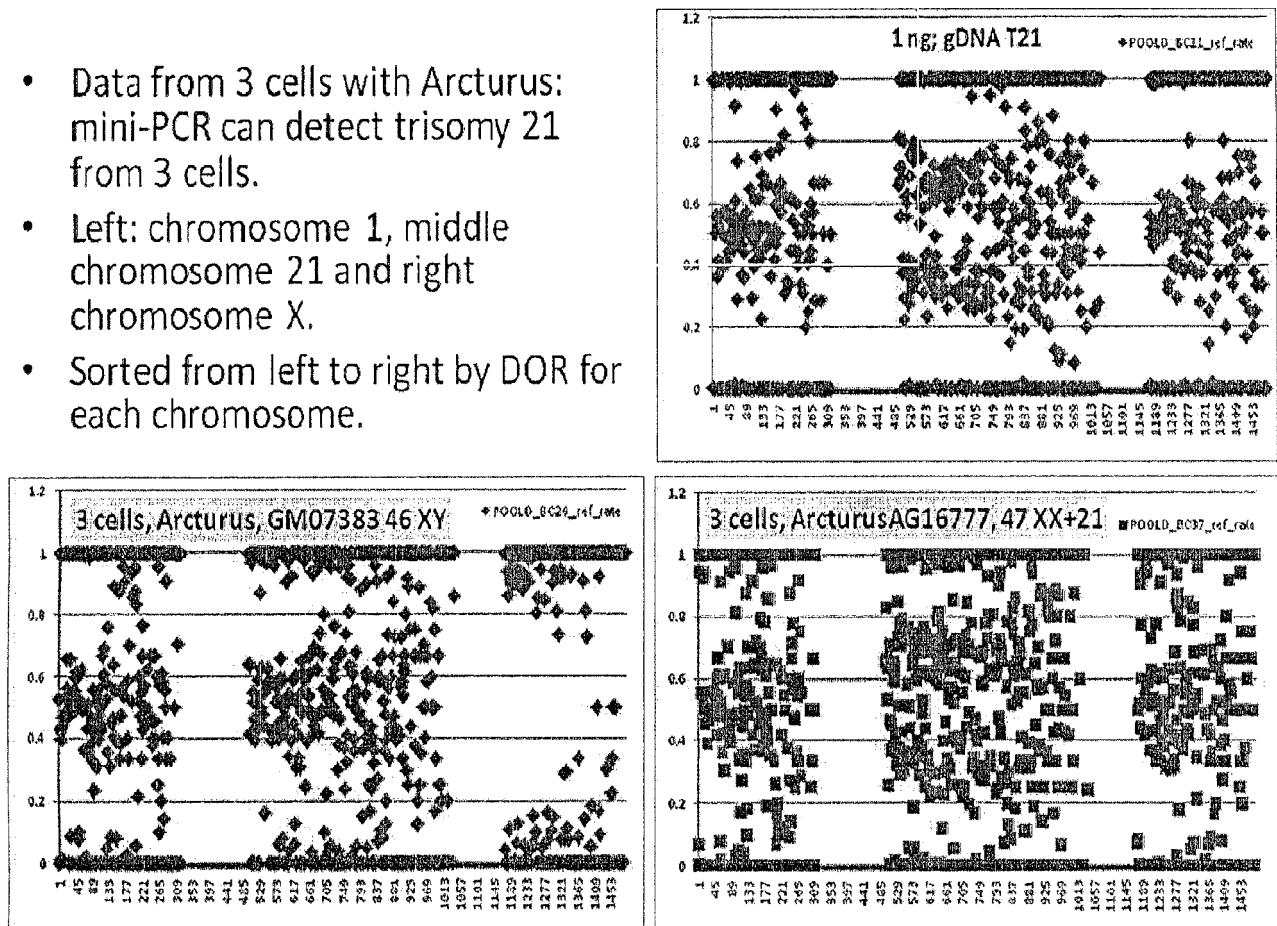


Figure 25

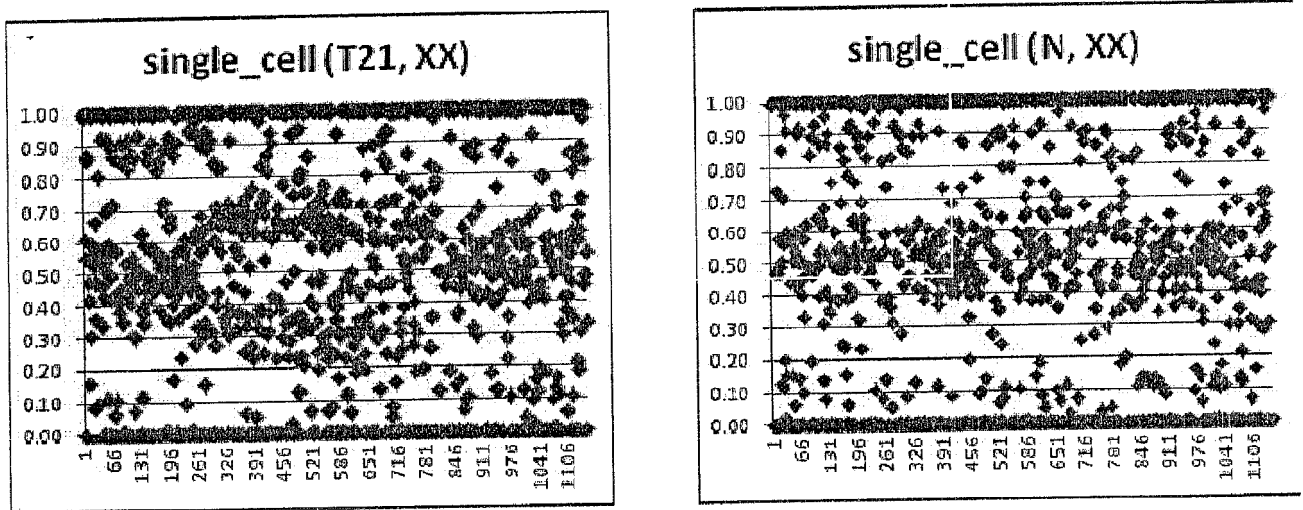


Figure 26

Figure 27

Minor Allele Frequencies for NIPD Targets, Chromosome 21

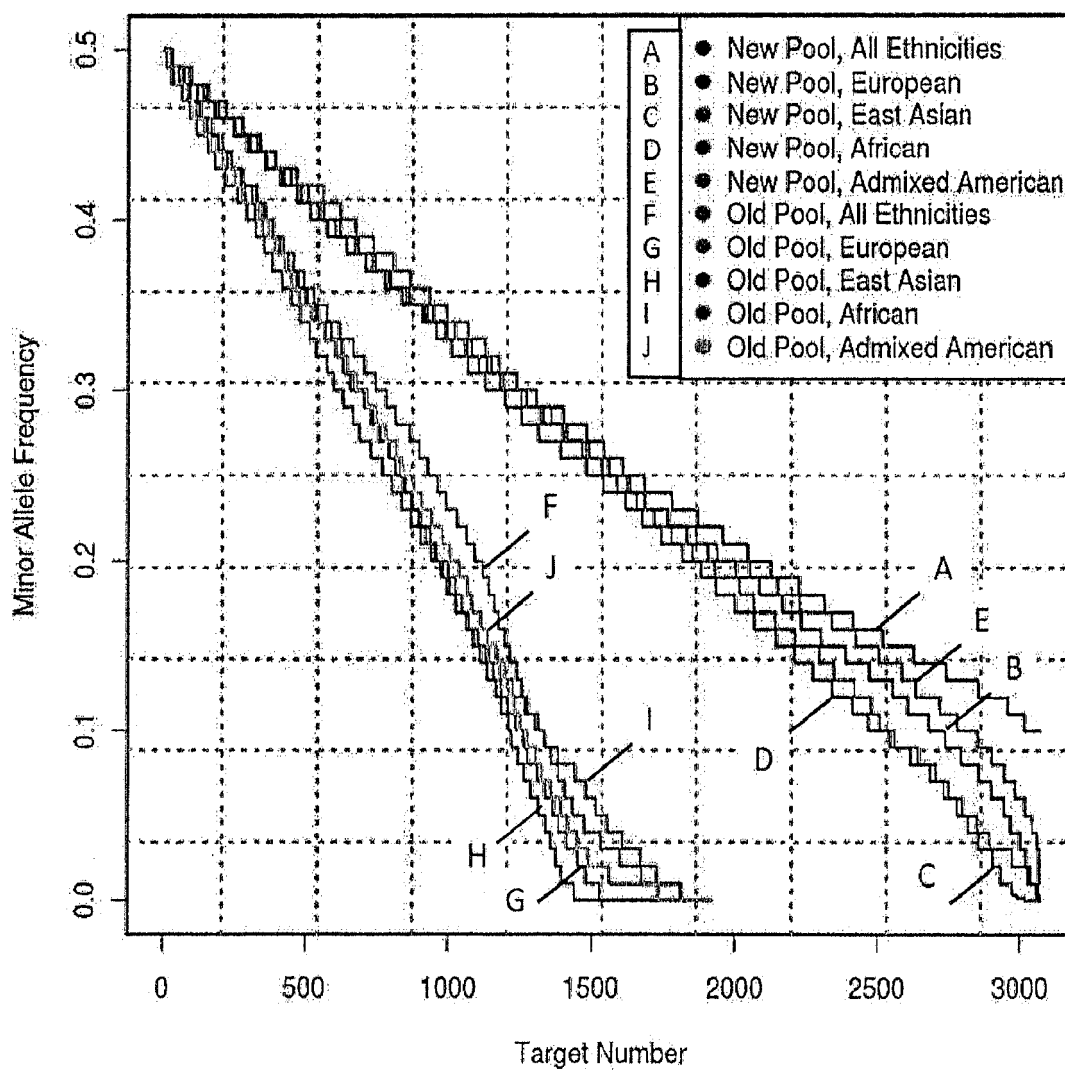
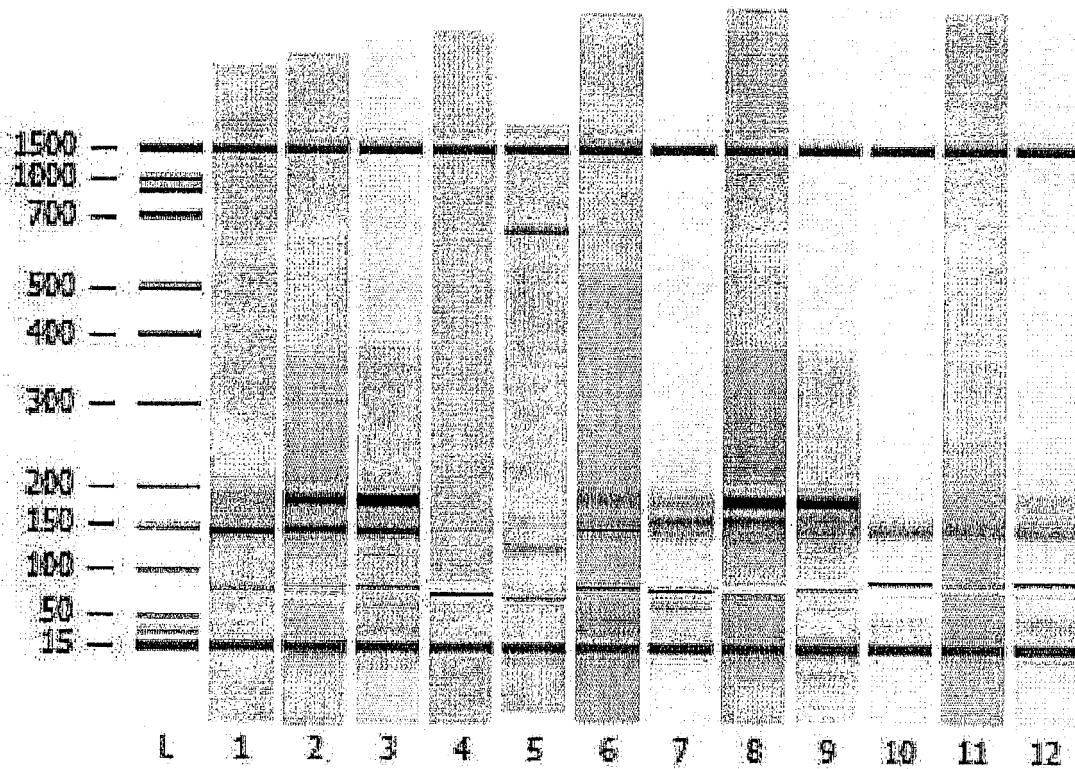


Figure 28A



Direct PCR 2.4k 24ng 10nM

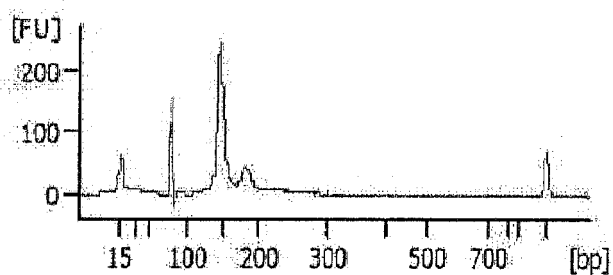


Figure 28B

Direct PCR 2.4k 80ng 10nM

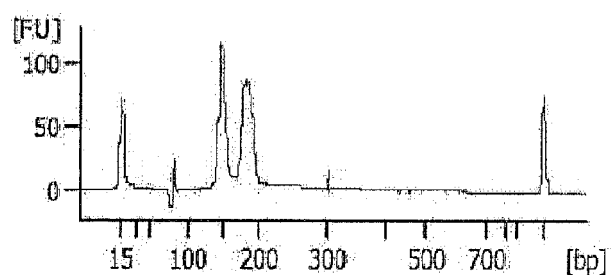


Figure 28C

Direct PCR 2.4k 250ng 10nM

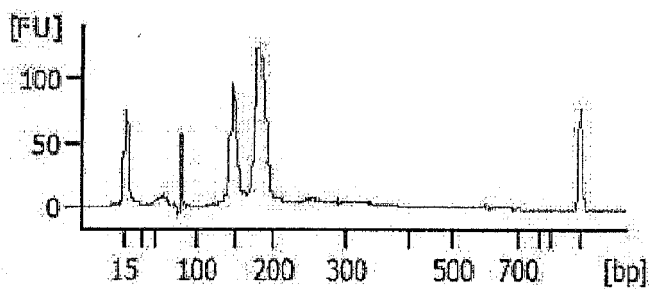


Figure 28D

Direct PCR 2.4k 24ng 1nM

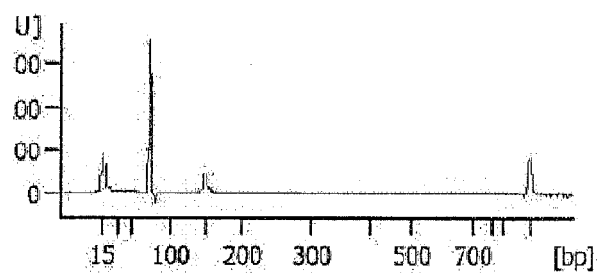


Figure 28E

Direct PCR 2.4k 80ng 1nM

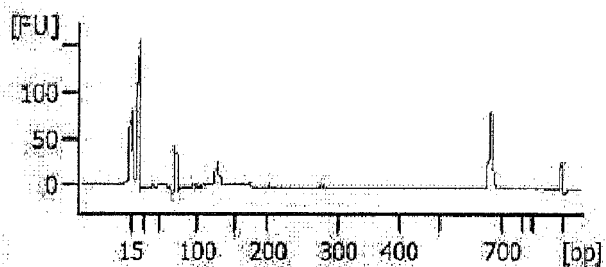


Figure 28F

Direct PCR 2.4k 250ng 1nM

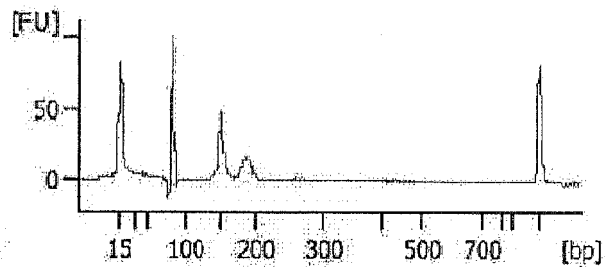


Figure 28G

Direct PCR 20k 24ng 10nM

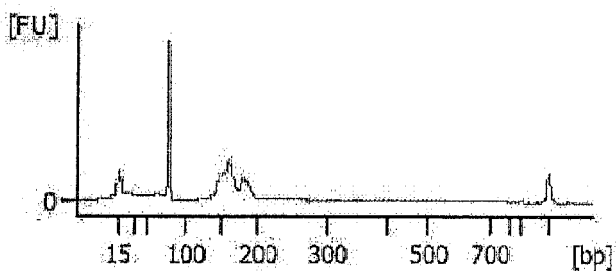


Figure 28H

Direct PCR 20k 80ng 10nM

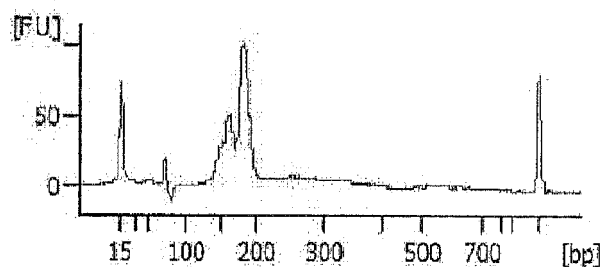


Figure 28I

Direct PCR 20k 250ng 10nM

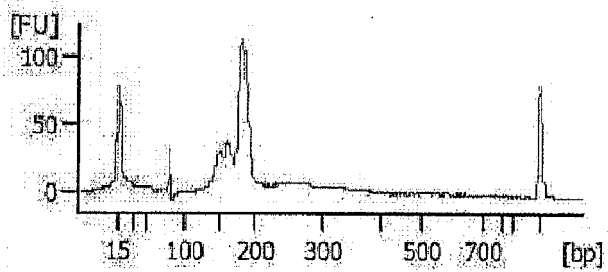


Figure 28J

Direct PCR 20k 24ng 1nM

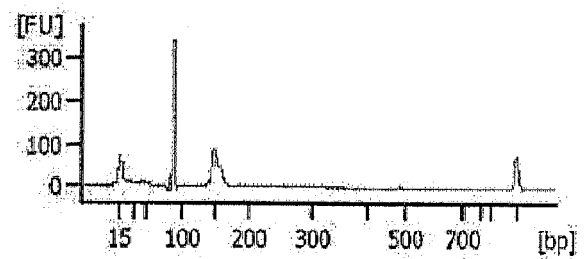


Figure 28K

Direct PCR 20k 80ng 1nM

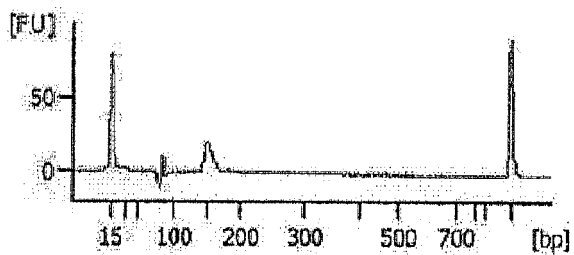


Figure 28L

Direct PCR 20k 250ng 1nM

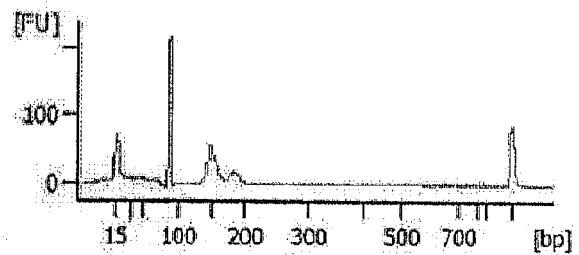
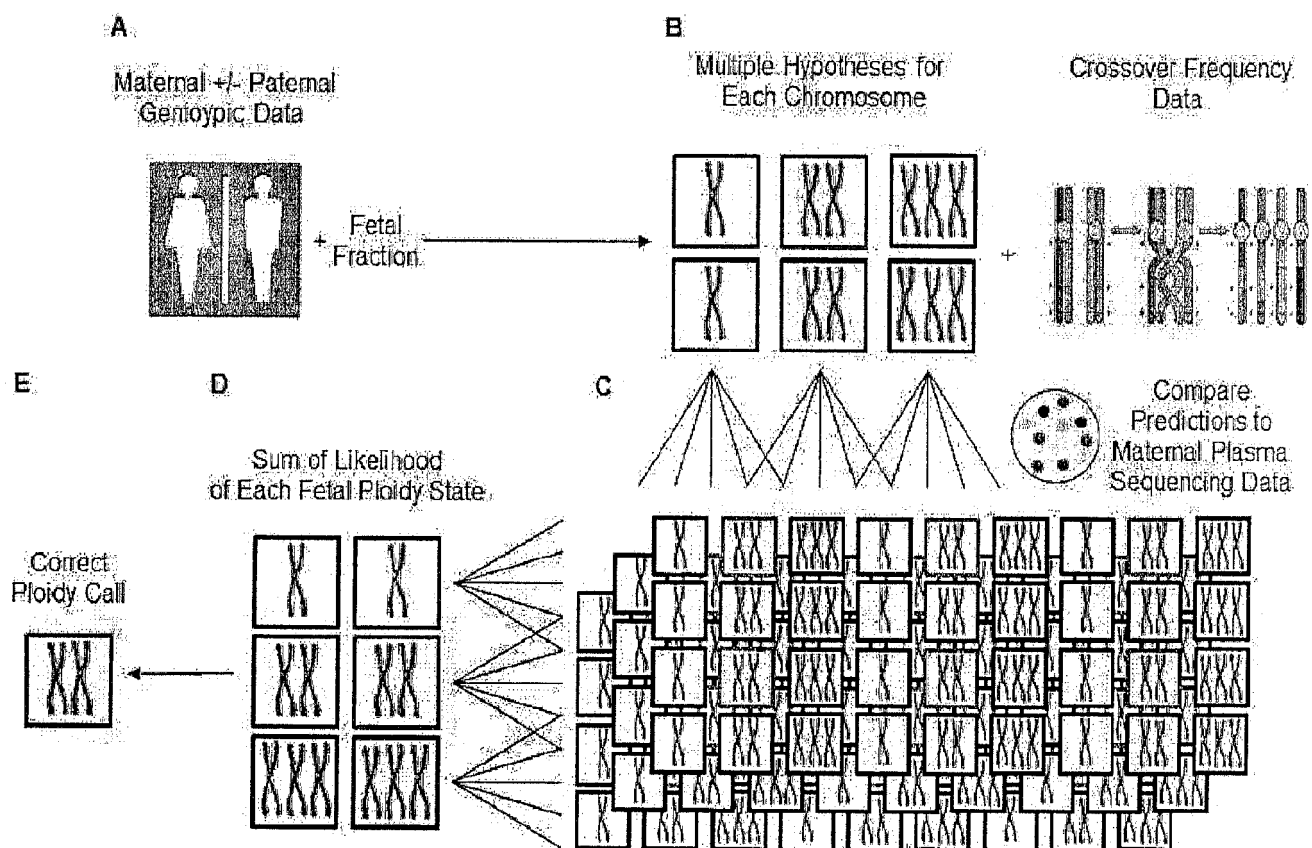
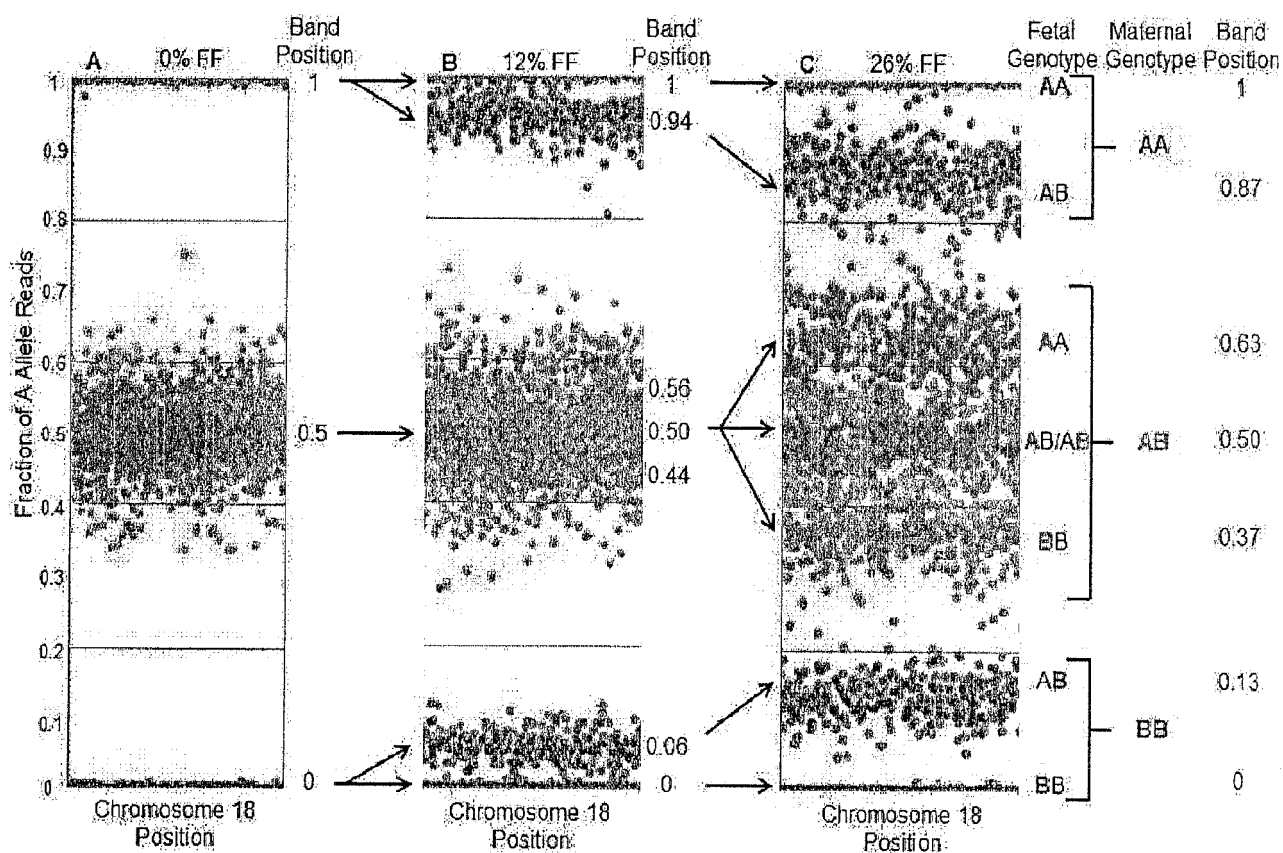


Figure 28M

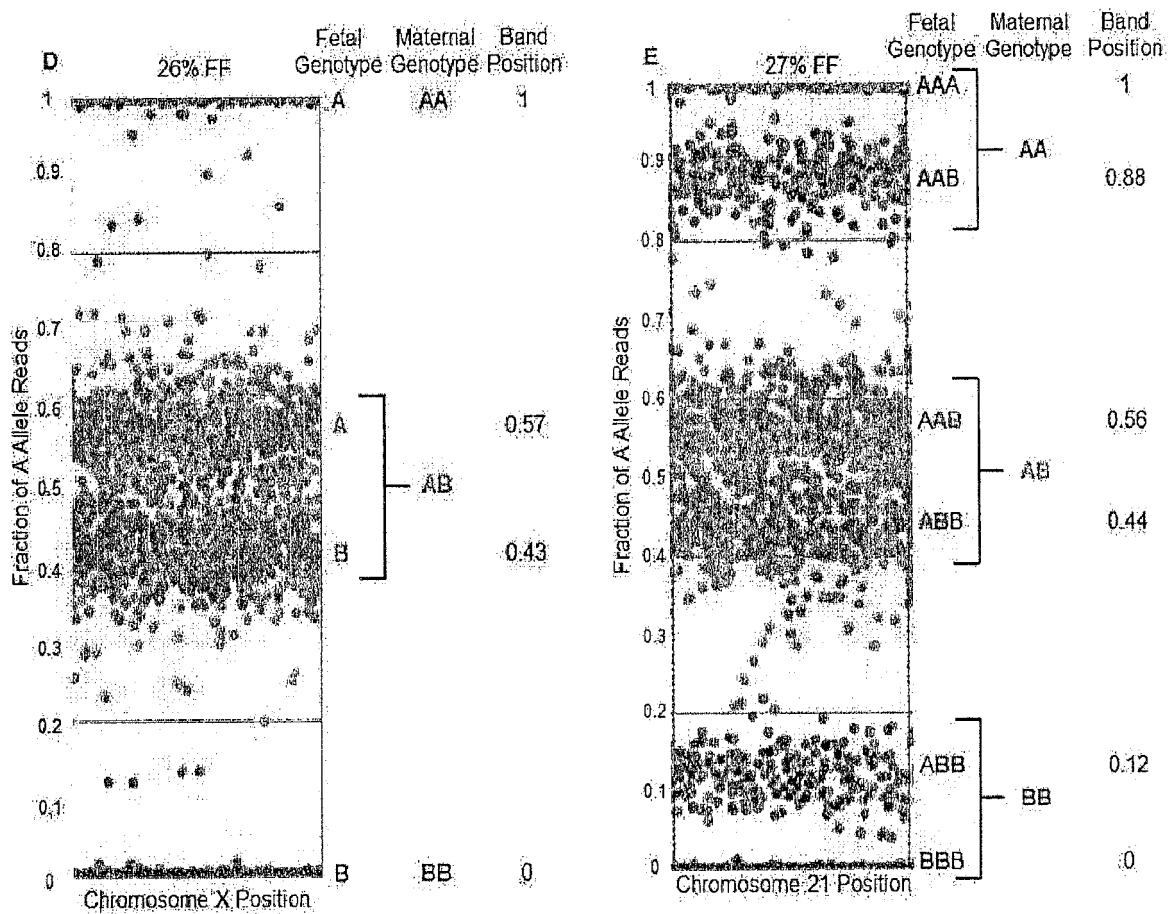
Figures 29A-29E



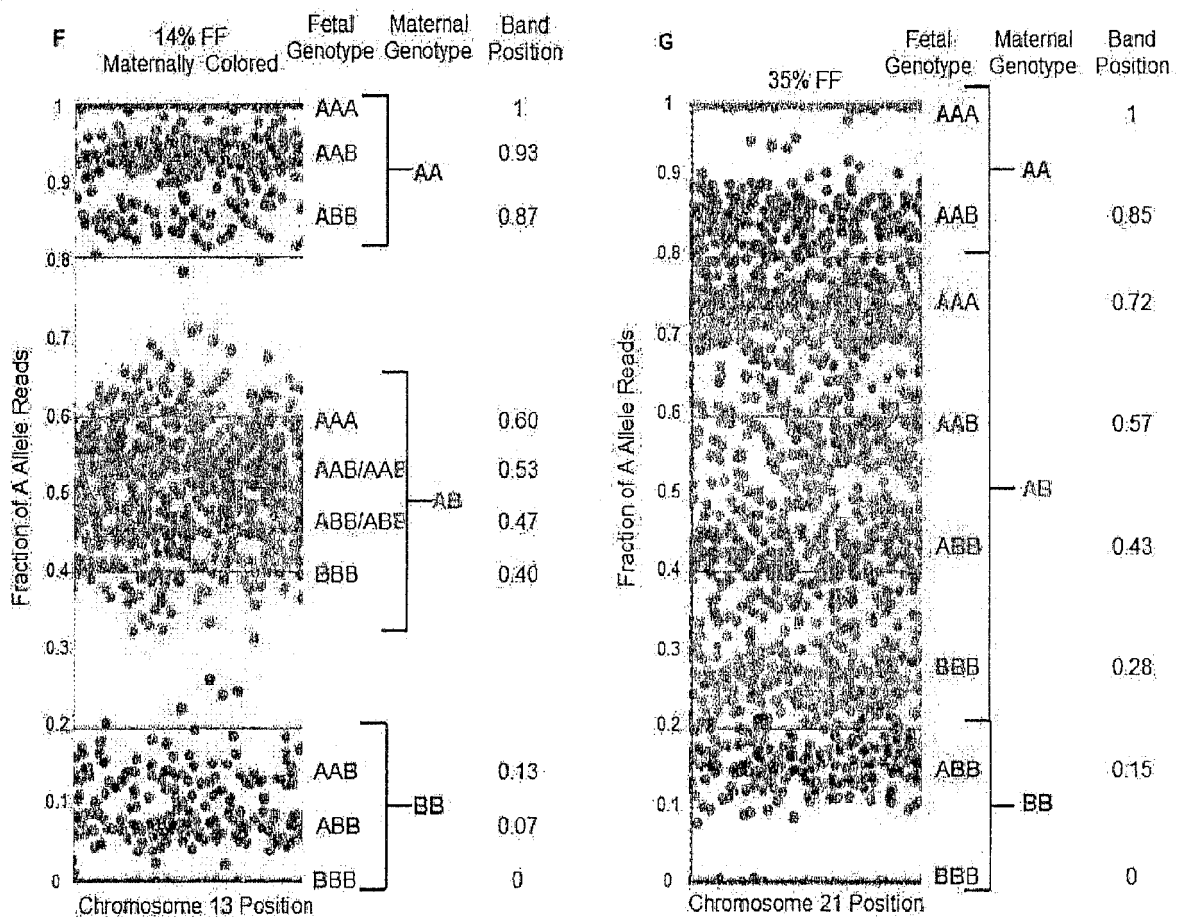
Figures 30A-30C



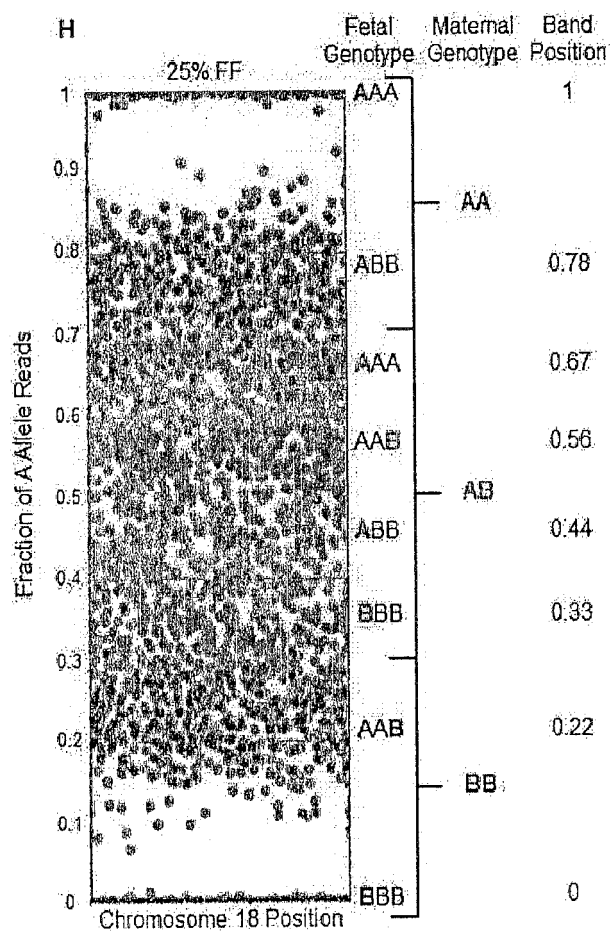
Figures 30D and 30E



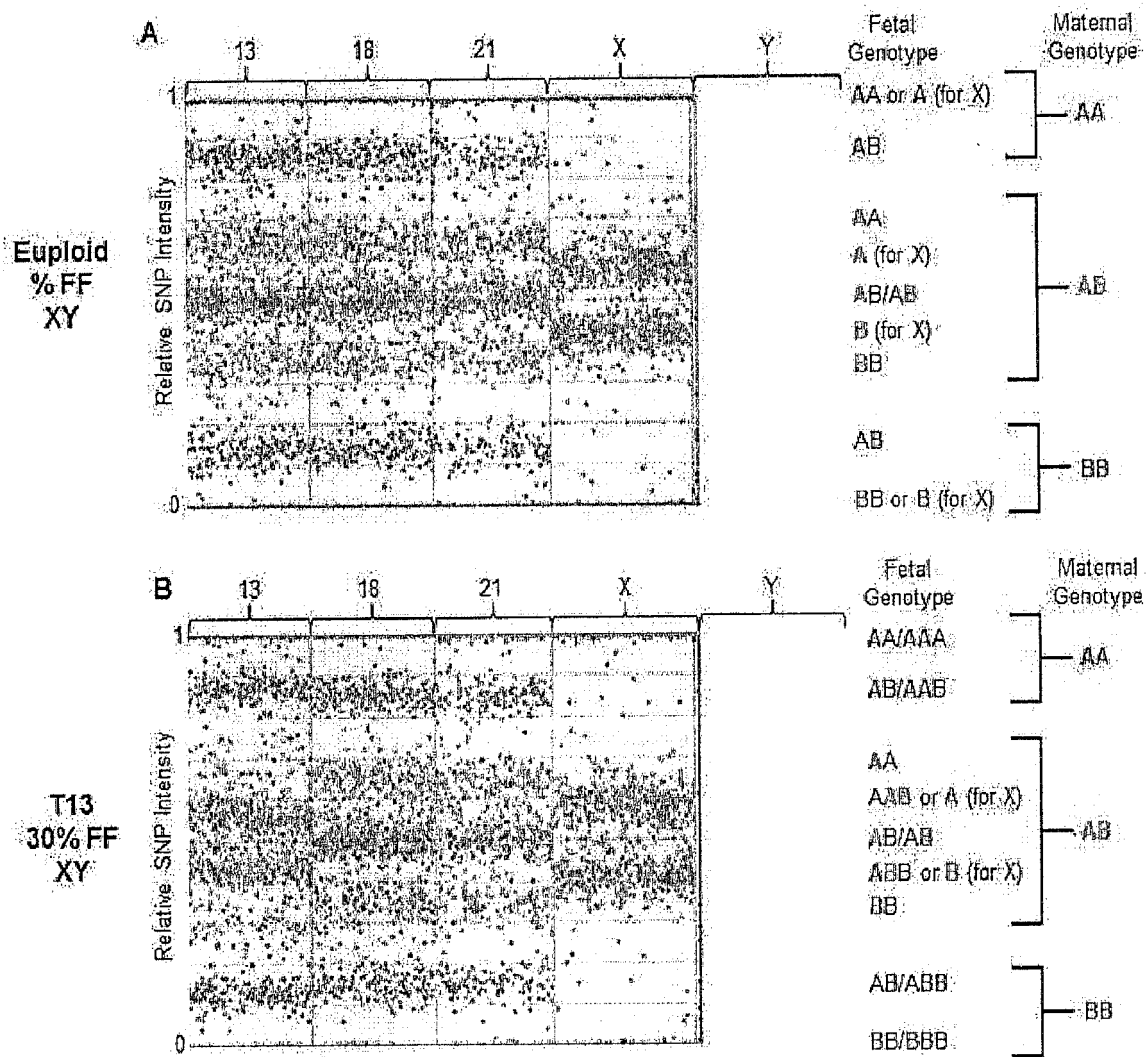
Figures 30F and 30G



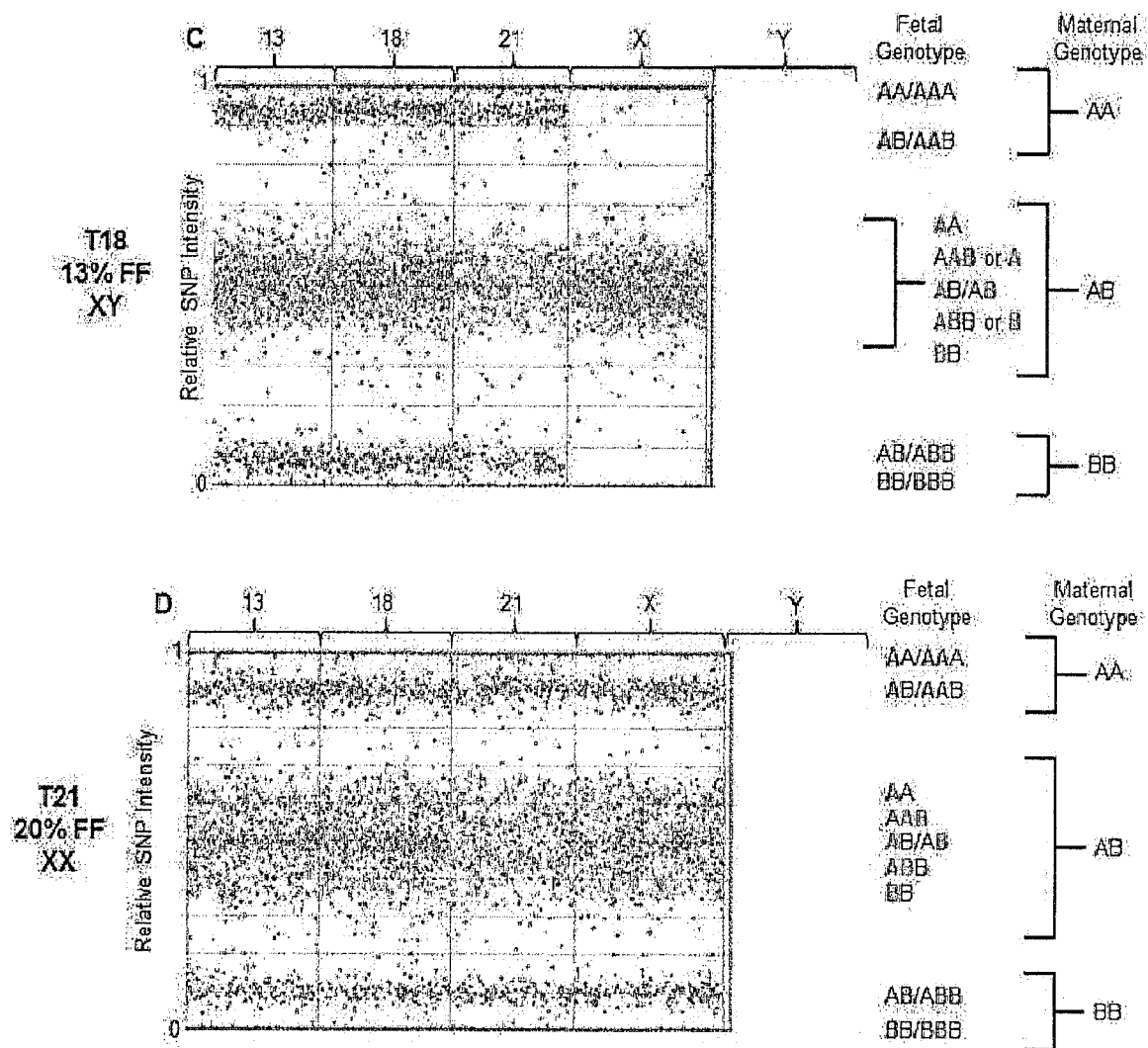
Figures 30H



Figures 31A and 31B



Figures 31C and 31D



Figures 31E and 31F

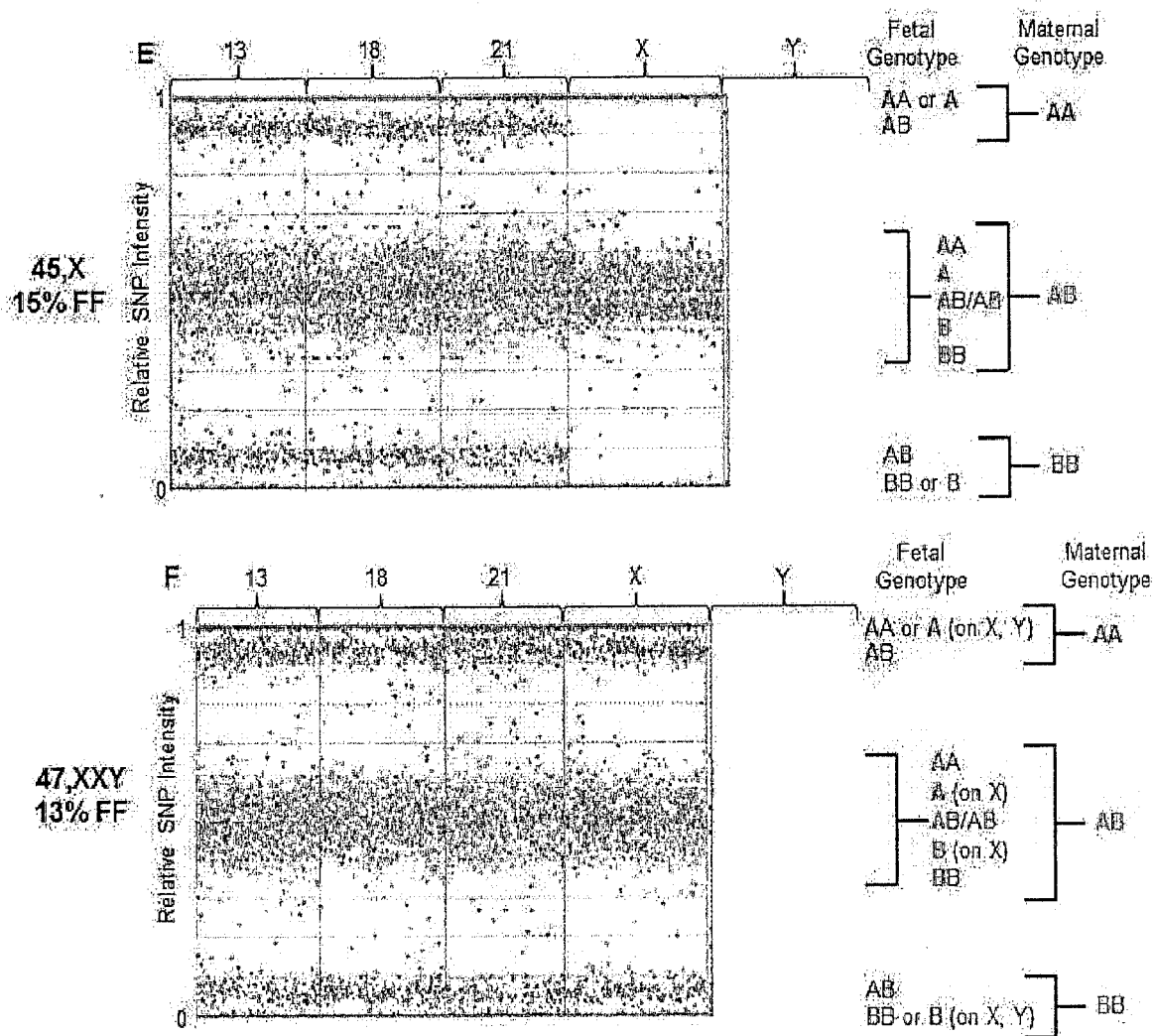


Figure 31G

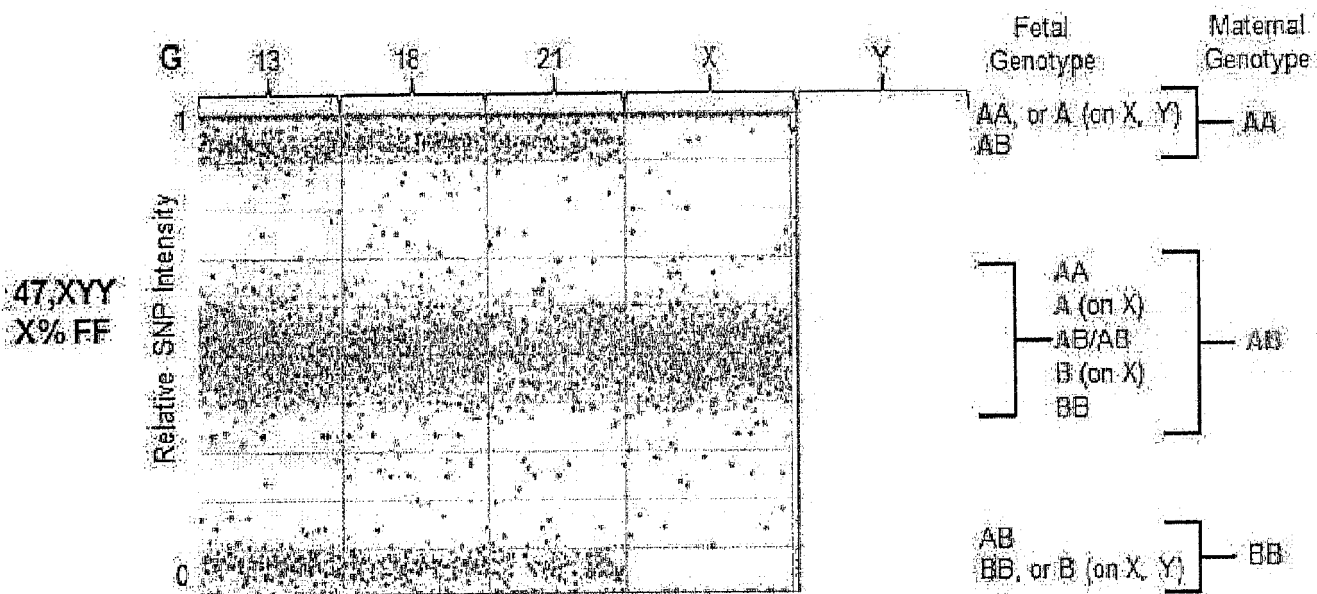


Figure 32

