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Genetic analysis for a prenatal diagnosis of complex chromosomal rearrangement

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Objective

To investigate the chromosomal aberration of a fetal that was implicated to have sex chromosome anomalies from noninvasive prenatal detection, and then provide accurate genetic counseling for the pregnant women.

Methods

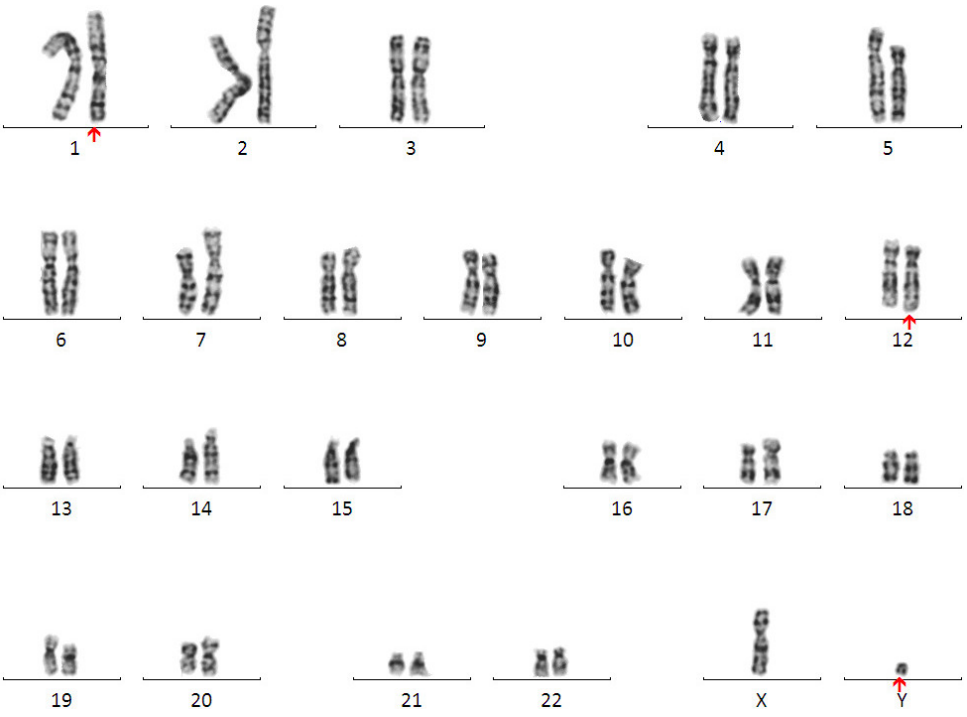
Four-dimensional ultrasonography was performed in combination with chromosome karyotype analysis, single nucleotide polymorphism array (SNP array), and multiplex real-time PCR analysis for Y chrommosome mircodeletions.

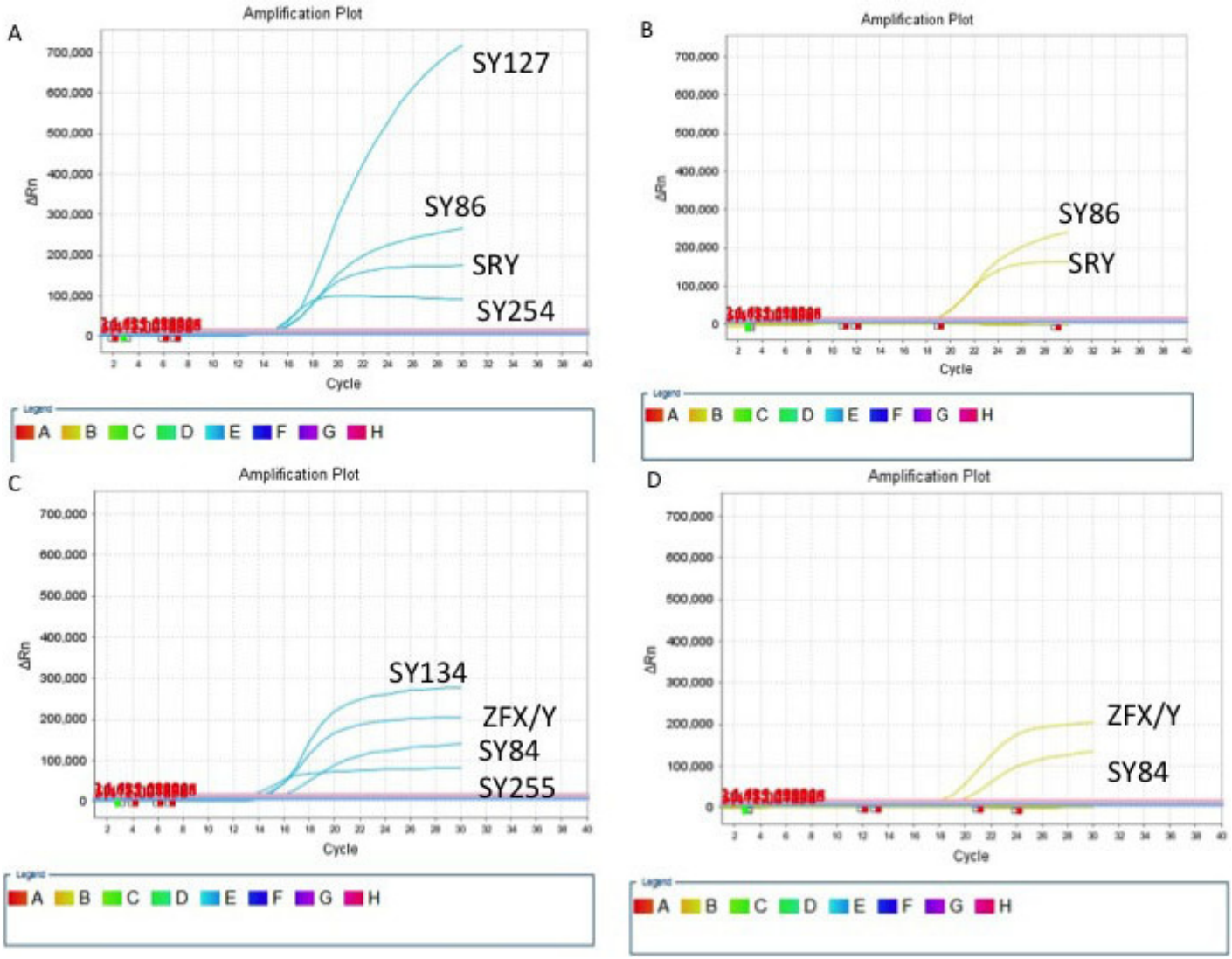
Results

The chromosome karyotype is 46, X, del(Y)(q?), t(1;12)(q32;q24. 1)dn; The results of SNP array is arr[19] Yq11. 221q11. 23 (16221460-28799654), ~ 12. 5Mb deletion. Analysis of Y chrommosome mircodeletions shows that 4 STS loci in the AZFb and AZFc regions are all deleted, further verifying the Y chromosome microdeletions. Doppler ultrasound examination showed no apparent abnormal changes in fetal structure.

Conclusion

The fetus carries a translocation between chromosome 1 and 12, and a partial Y chromosome deletion. Usually, the fetus will be clinically normal after birth, but developments fertility disorder at adulthood. Thus, four-dimensional ultrasonography in combination with multiple genetic testing provides technical support for the prevention of birth defects. Figure legends: Figure 1: Karyotype analysis Figure 2: Genomic copy number variation analysis shows mircodeletions on Y chromosome Figure 3: Multiplex real-time PCR analysis shows microdeletions on 4 STS loci in the AZFb and AZFc regions of Y chromosome.

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Amplification Plot
SY127
SY86
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SY254
ΔRn
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B
Amplification Plot
SY86
SRY
ΔRn
Cycle
C
Amplification Plot
SY134
ZFX/Y
SY84
SY255
ΔRn
Cycle
D
Amplification Plot
ZFX/Y
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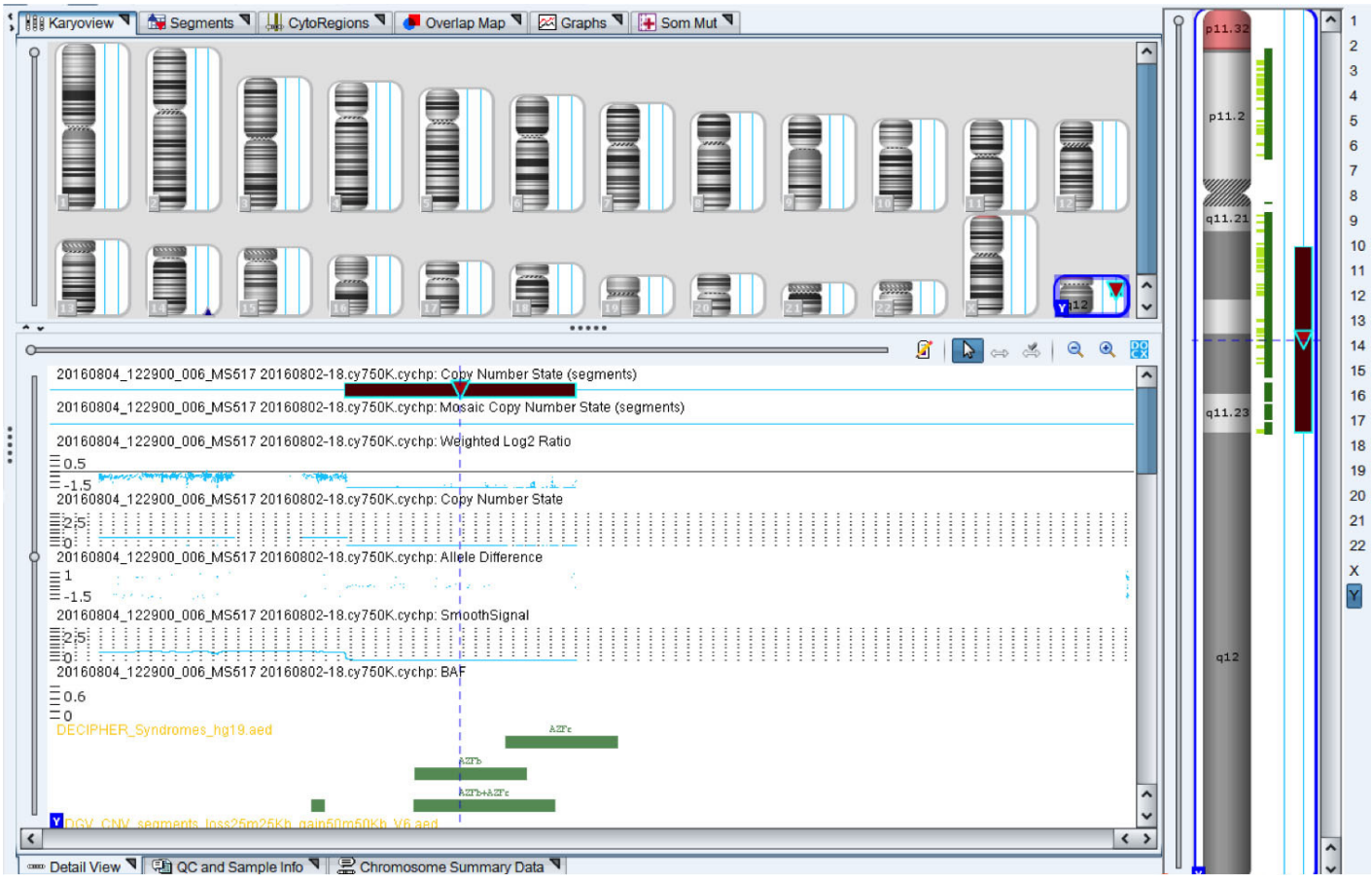
A B C D E F G H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Detail View
QC and Sample Info
Chromosome Summary Data