Cobioer Gene Technology Co., Ltd.

BACKGROUND

Noninvasive prenatal testing (NIPT) is increasingly used for prenatal screening.NIPT, which is characterized by non-invasiveness and accuracy, is widely used for prenatal screening. NIPT has been mainly used to screen for common fetal chromosome aneuploidies, including trisomy 21 syndrome (T21), trisomy 18 syndrome (T18), and trisomy 13 syndrome (T13).

In recent years, NIPT has also been used to screen for sex chromosome aneuploidy (SCA) as more in-depth studies have been reported.

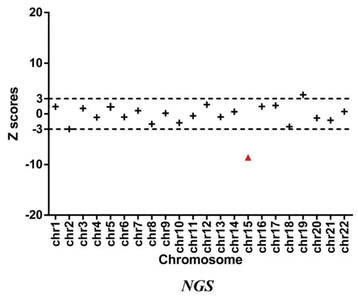
At present, due to the improvement of library construction technology and the reduction of sequencing cost, the detection range of NIPT is gradually expanded, and the detection of MMS or CNV is gradually becoming possible.

INTRODUCTION

CB-Gene has launched NIPT Reference Sandards, including autosomal non-integer copy number variations, sex chromosome non-integer copy number variations, and microdeletion/microduplication reference standards.

The process quality control for sample extraction, library preparation, and on-machine sequencing of Non-Invasive Prenatal Testing (NIPT) kits can be effectively monitored.

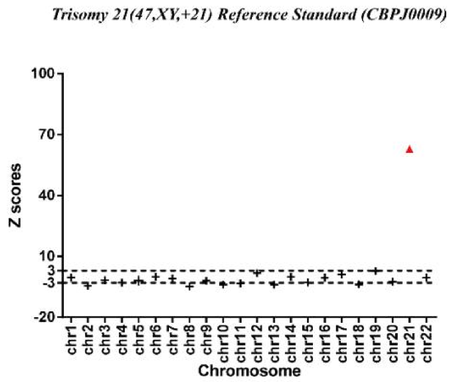
Prader-Willi syndrome (46,XY,del(15)(q11.2q13)) Reference Standard (CBPJ0007)



These reference standards are suitable for detecting cell-free DNA (cfDNA) in maternal peripheral blood using genome-wide methods, such as Next-Generation Sequencing (NGS).

PRODUCT DATA

Trisomy 21(47,XY,+21) Reference Standard (CBPJ0009)



Trisomy 13 (47,XY,+13) Reference Standard (CBPJ0010)

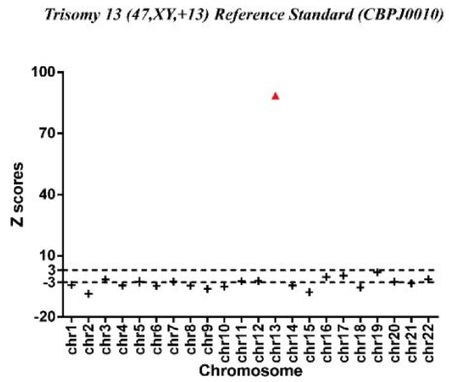
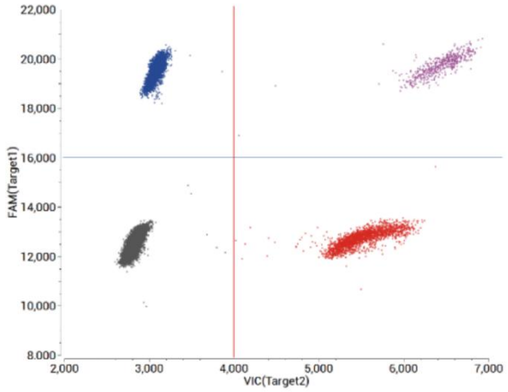


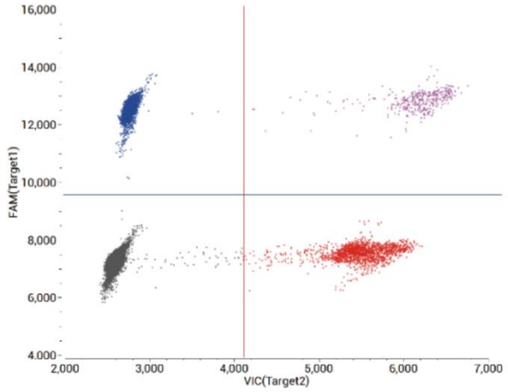
Fig 1. Results of NGS with Trisomy 21 (47,XY,+21) Reference Standard and Trisomy 13 (47,XY,+13) Reference Standard.

Trisomy 21(47,XY,+21) Reference Standard (CBPJ0009)



CNV=3.01

Trisomy 13 (47,XY,+13) Reference Standard (CBPJ0010)



CNV=3.07

Fig 2. Results of ddPCR with Trisomy 21 (47,XY,+21) Reference Standard and Trisomy 13 (47,XY,+13) Reference Standard.

NIPT reference	Product	Cat.No
Autosomal non-integer copy number variations	Trisomy 13 (47,XY,+13) Reference Standard	CBPJ0010
	Trisomy 18(47,XX,+18) Reference Standard	CBPJ0002
	Trisomy 21 (47,XY,+21) Reference Standard	CBPJ0009
	Trisomy 21(47,XX,+21) Reference Standard	CBPJ0001
	Trisomy 9 (47,XY,+9) Reference Standard	CBPJ0014
Sex chromosome non-integer copy number	Klinefelter Syndrome (47，XXY) Reference Standard	CBPJ0005
Microdeletion/microduplication reference standards	11q23.3 del (46,XX,del(11)(q23.3)) Reference Standard	CBPJ0015
	18P-syndrome (46,XX,del(18)(p11.2)) Reference Standard	CBPJ0008
	18Q-syndrome (46,XX,del(18)(q22)) Reference Standard	CBPJ0013
	Angelman syndrome (46,XX,del(15)(q11q13)) Reference Standard	CBPJ0006
	DiGeorge syndrome (46,XX,del(22)(q11)) Reference Standard	CBPJ0011
	Prader-Willi syndrome (46,XY,del(15)(q11.2q13)) Reference Standard	CBPJ0007
	Trisomy 9 (47,XY,+9,del(9)(q11)) Reference Standard	CBPJ0003

TEST PRINCIPLE

For chromosomal non-integer copy number abnormal samples, the abnormal chromosome is identified through NGS. Based on the human genome sequence, primers and probes targeting the relevant abnormal chromosomes are designed. The ddPCR method is then used for further validation, and the chromosomal status is determined by analyzing the CNV value.

Microdeletion and microduplication reference standards are validated using both NGS and CMA methods. NGS is used to identify the abnormal chromosomes, while CMA is employed to pinpoint the specific abnormal regions in the samples.

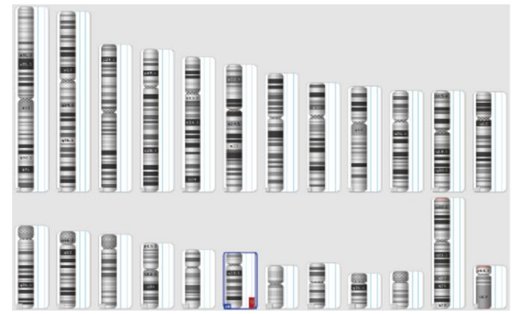


Fig 3. Results of CMA with 18Q-syndrome (46,XX,del(18)(q22)) Reference Standard.

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