18P-syndrome (46,XX,del(18)(p11.2)) Reference Standard

CBPJ0008

I. Description

NIPT reference products use gDNA from human cell lines as raw materials, which are fragmented and then added to human plasma to simulate real clinical samples. They can monitor the quality control of sample extraction, library preparation and sequencing of non-invasive prenatal testing (NIPT) kits. They are suitable for the detection of cell-free DNA (cfDNA) in peripheral blood of pregnant women using the whole genome method (massive-parallel sequencing MPS).

CB-Gene NIPT reference products can be customized for common chromosomal abnormalities: aneuploidy of four common chromosomes (9/21/13/18); aneuploidy of one sex chromosomes is aneuploid (XXY/47) ; Common microdeletion and microduplication syndrome types,such as DGS,AS,PWS,11q23.3 del, etc.

II. General information

Name	18P-syndrome (46,XX,del(18)(p11.2)) Reference Standard
Cat. No.	CBPJ0008
Format	Genomic DNA
Unit Size	1ug
Intended Use	Research Use Only
Buffer	Tris-EDTA
Storage Conditions	2-8℃
Expiry	36 months from the date of manufacture

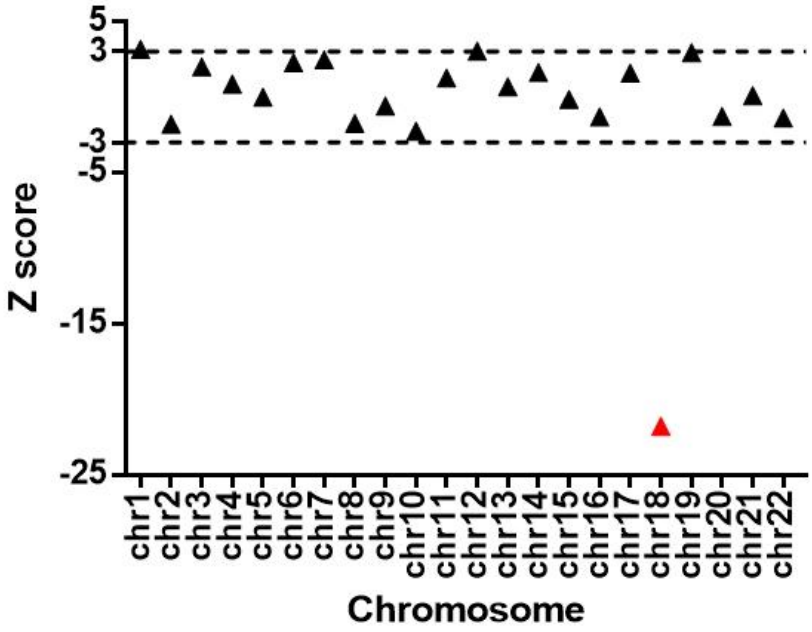
III.Technical Data

Mutation	18P-syndrome
Karyotype	46,XX,del(18)(p11.2)
Chromosome (GRCh37) abnormal region (CMA)	chr18:136,228-15,170,636
Mutation Type	Loss
Length (bp)	15034409
ACMG variant classification	Pathogenicity

IV.Representative Data

Z-Score：

chr	Z-Score	chr	Z-Score
chr1	3.16	chr12	3.04
chr2	-1.79	chrl3	0.68
chr3	2	chr14	1.61
chr4	0.84	chr15	-0.15
chr5	-0.02	chr16	-1.33
chr6	2.26	chr17	1.57
chr7	2.46	chr18	-21.78
chr8	-1.75	chr19	2.94
chr9	-0.62	chr20	-1.3
chr10	-2.27	chr21	0.09
chr11	1.26	chr22	-1.41



Chromosome simulated karyotype：