# Angelman syndrome (46,XX,del(15)(q11q13)) Reference Standard

CBPJ0006

## 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 | Angelman syndrome (46,XX,del(15)(q11q13)) Reference Standard |
| Cat. No. | CBPJ0006 |
| 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 |

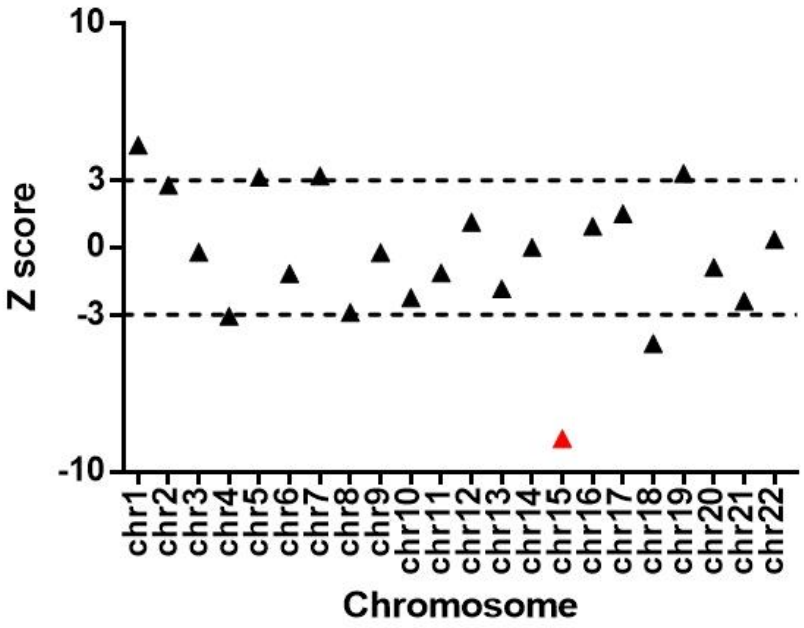
## III.Technical Data

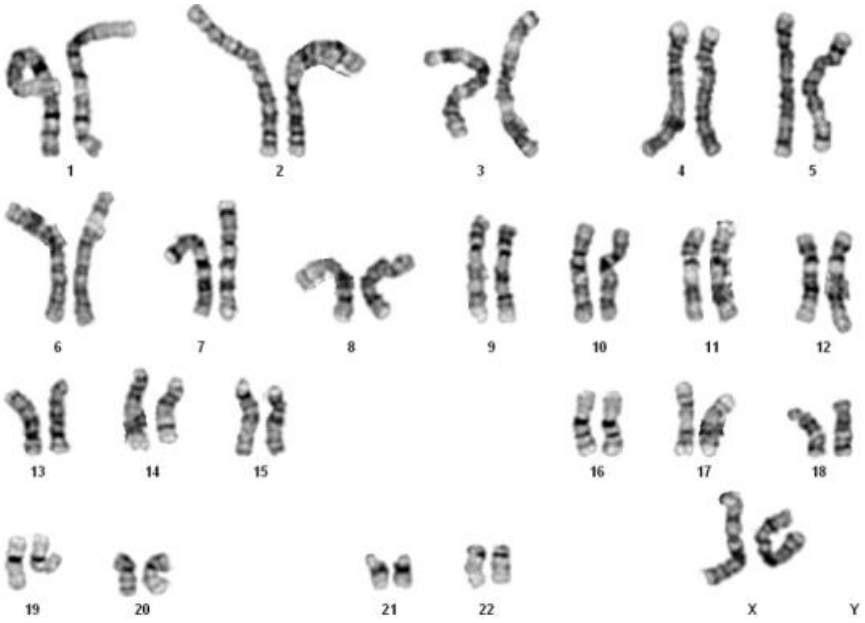
| | |
|---|---|
| Mutation | Angelman syndrome |
| Karyotype | 46,XX,del(15)(q11q13) |
| Chromosome (GRCh37) abnormal region (CMA) | chr15:22,770,422-28,644,578 |
| Mutation Type | Loss |
| Length (bp) | 5874157 |
| ACMG variant classification | Pathogenicity |

## IV.Representative Data

Z-Score：

| chr | Z-Score | chr | Z-Score |
|---|---|---|---|
| chr1 | 4.56 | chr12 | 1.13 |
| chr2 | 2.78 | chr13 | -1.84 |
| chr3 | -0.21 | chr14 | 0.01 |
| chr4 | -3.05 | chr15 | -8.52 |
| chr5 | 3.15 | chr16 | 0.96 |
| chr6 | -1.16 | chr17 | 1.51 |
| chr7 | 3.19 | chr18 | -4.28 |
| chr8 | -2.9 | chr19 | 3.31 |
| chr9 | -0.23 | chr20 | -0.88 |
| chr10 | -2.23 | chr21 | -2.38 |
| chr11 | -1.14 | chr22 | 0.35 |


10
3
0
-3
-10
Z score
chr1
chr2
chr3
chr4
chr5
chr6
chr7
chr8
chr9
chr10
chr11
chr12
chr13
chr14
chr15
chr16
chr17
chr18
chr19
chr20
chr21
chr22
Chromosome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X
Y

Chromosome simulated karyotype: