Angelman syndrome (46,XY,del(15)(q11.2q13.1)) Reference Standard

CBPJ0017

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,XY,del(15)(q11.2q13.1)) Reference Standard
Cat. No.	CBPJ0017
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,XY,del(15)(q11.2q13.1)
Chromosome (GRCh37) abnormal region (CMA)	chr15:23632678-28534458
Mutation Type	Loss
Length (bp)	4901781
ACMG variant classification	Pathogenicity

IV.Representative Data

Chromosome simulated karyotype：