Trisomy 21 (Down Syndrome)

(Core Condition)

Description

The presence of three copies of all or a large part of chromosome 21.

Inclusions

Down syndrome
Mosaic Down syndrome
Mosaic trisomy 21
Translocation Down syndrome
Translocation trisomy 21
Trisomy 21, not otherwise specified

Exclusions

Balanced translocations involving chromosome 21
"Downs facies" without associated trisomy 21.

ICD-9-CM Codes

758.0

ICD-10-CM Codes

Q90.0 – Q90.9

CDC/BPA Codes

758.00 – 758.09

Diagnostic Methods

Down syndrome may be suspected on physical examination. However, it may be diagnosed conclusively only through direct analysis of the infant's chromosomes (karyotype). The chromosomes may be obtained from blood or tissue cells.

Prenatal Diagnoses Not Confirmed Postnatally

Down syndrome may be included when only diagnosed through direct analysis of fetal chromosomes or molecular cytogenetic analysis (typically chromosomal microarray or fluorescence in situ hybridization) of cells obtained from amniocentesis, chorionic villus sampling (CVS), or percutaneous umbilical blood sampling (PUBS). However, when mosaic trisomy 21 is noted, the defect should be confirmed postnatally on a specimen obtained directly from the infant or fetus after birth (see below).

Additional Information:

When the two copies of chromosome 21 from one parent do not separate during egg or sperm formation, three copies of the entire chromosome 21 will be present in the fetus. In this instance, the karyotype is written as 47,XX,+21 or 47,XY,+21. This is the most common type of trisomy 21 and is associated with advanced maternal age, particularly of 35 years or greater.

Translocation trisomy 21 occurs when two separate copies of chromosome 21 are present, but a third copy of part of chromosome 21 is attached to another chromosome. In this instance, there are 46 total