

DOWN
SYNDROME
[MEDICAL
BIOLOGY]

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DOWN SYNDROME:-

“Down’s syndrome causes a distinct facial appearance, intellectual disability and developmental delays. It may be associated with thyroid or heart disease”

Definition

- ◆ Down syndrome, also called Trisomy 21, is a genetic condition that causes delay in physical and intellectual development.
- ◆ There is extra genetic material from chromosome 21, so individuals with Down syndrome have 47 chromosomes in total instead of the usual 46.

TYPES OF DOWN SYNDROME

TRISOMY

THE EXTRA 21 CHROMOSOME PRODUCES A COMPLEMENT OF 47 CHROMOSOMES

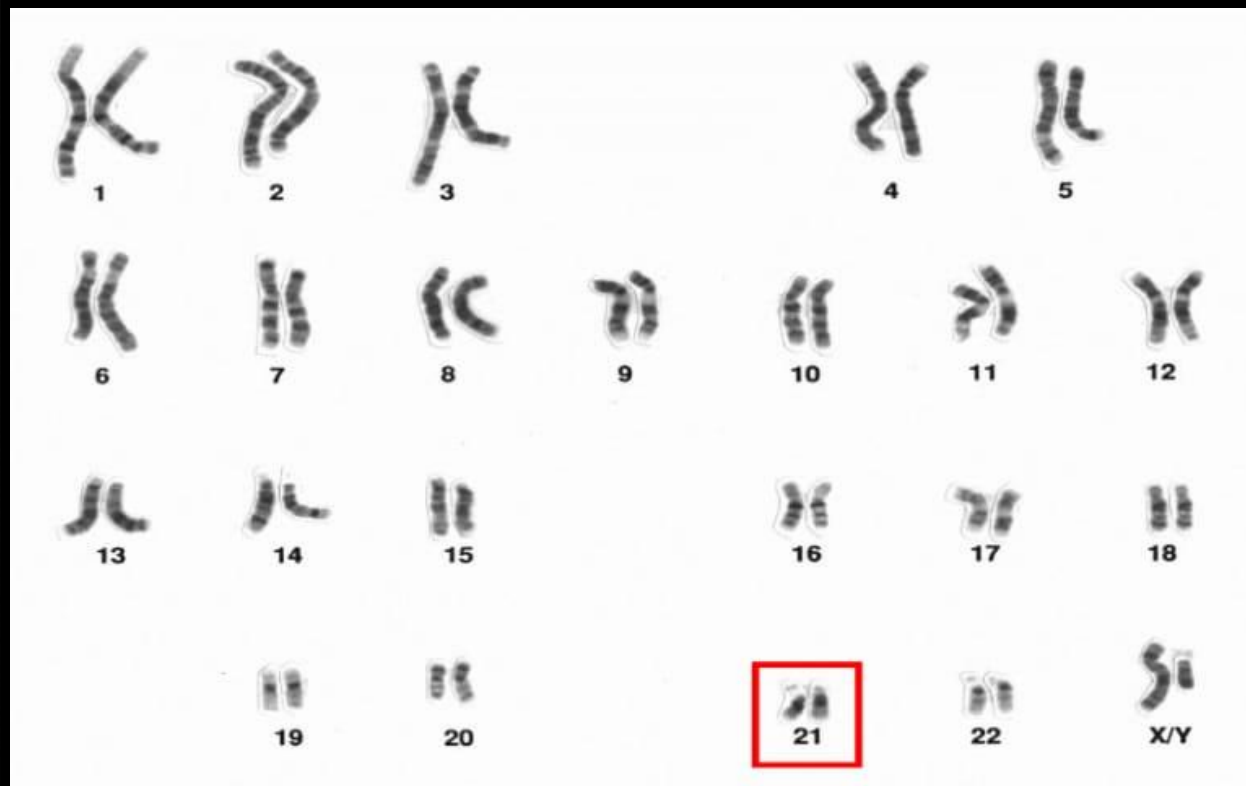
TRANSLOCATIO N

A SEGMENT OF A 21 CHROMOSOME IS FOUND ATTACHED TO OTHER PAIRS OF CHROMOSOMES

MOSIACISM

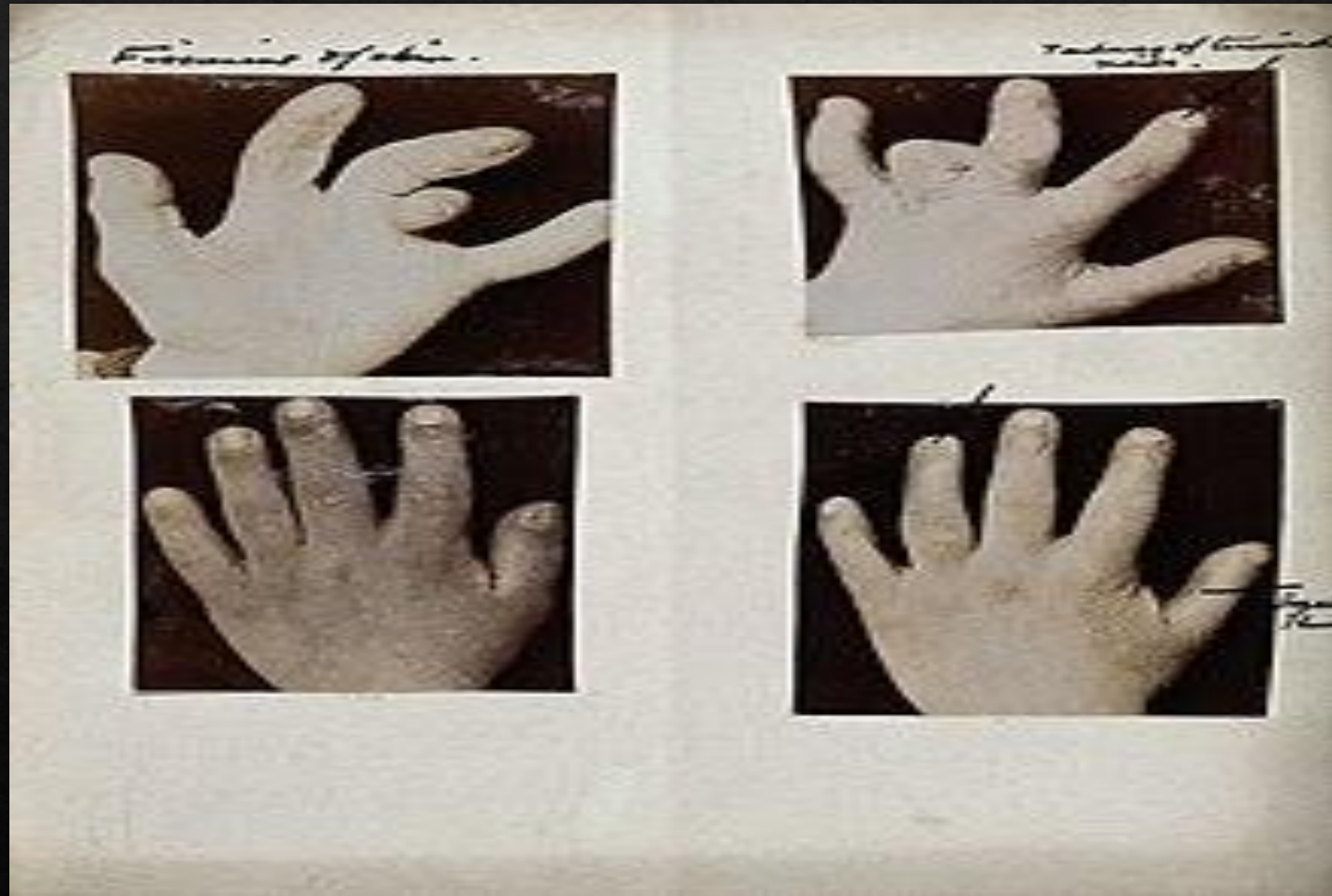
NONDISJUNCTION OCCURS AT A LARGER STAGE OF CELL DIVISION. THEREFORE, SOME CELLS HAVE THE NORMAL COMPLEMENT OF 46 CHROMOSOMES & OTHER CELLS 47 CHROMOSOMES.

KARYOTYPE OF DOWN SYNDROME

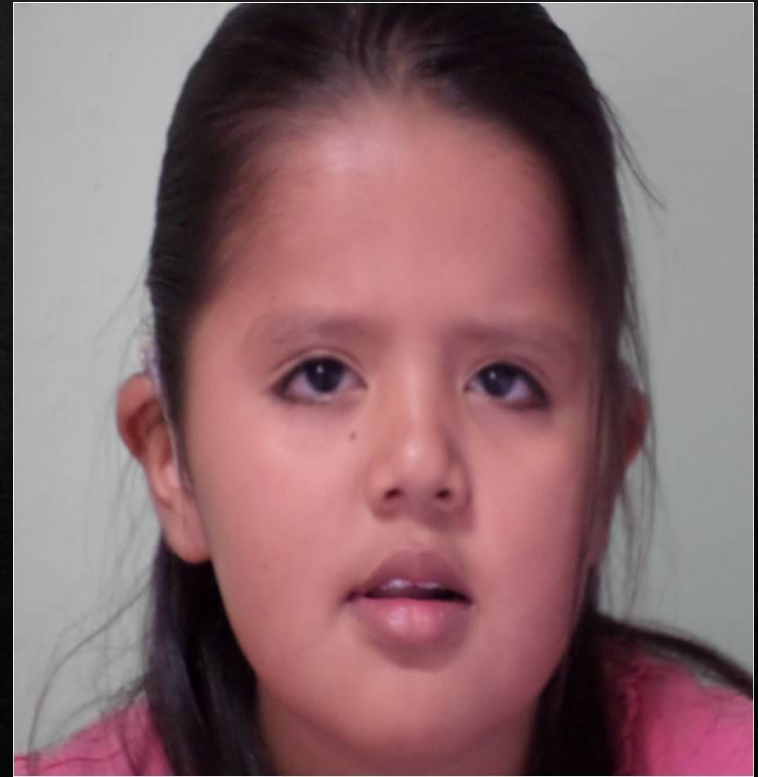


CLINICAL
MANIFESTATIO
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1] SMALL PINKY FINGERS THAT SOMETIMES
CURVE TOWARDS THE THUMB



2} FLAT FACE WITH UPWARD SLANT
TO THE EYE, SHORT & WIDE NECK.



3} BRUSH FIELD SPOTS

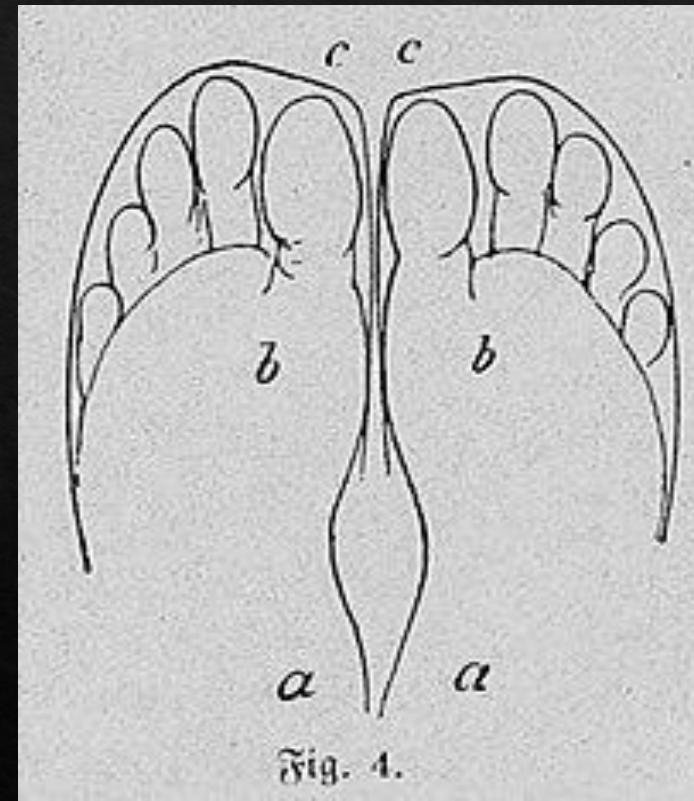


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4} MUSCLE HYPONTIA



5] EXCESSIVE SPACE BETWEEN LARGE TOE & SECOND TOE



COMPLICATIONS

- ◆ Delayed language, social and motor development.
- ◆ Growth retardation.
- ◆ Respiratory infections.
- ◆ Vision problems.
- ◆ Cataracts.
- ◆ Hearing loss.
- ◆ Increased risk for dementia.

DIAGNOSTIC TEST

1] CHRONIC VILLUS SAMPLING

- ◆ SAMPLE OF PLACENTA TAKEN EITHER THROUGH THE CERVIX OR THROUGH A NEEDLE INSERTED THROUGH THE ABDOMEN.
- ◆ TEST IS PERFORMED BETWEEN 8 TO 12 WEEKS.

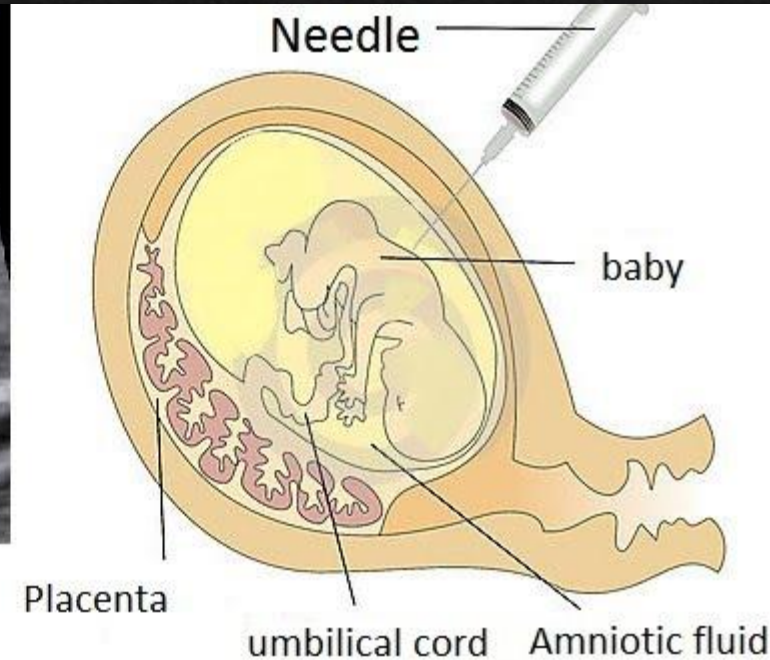
2] AMNIOCENTESIS

- ◆ REMOVAL OF A SMALL AMOUNT OF AMNOTIC FLUID THROUGH A NEEDLE INSERTED IN THE ABDOMEN. CELLS ARE ANALYZED FOR THE PRESENCE OF CHROMOSOMAL ABNORMALITIES.
- ◆ TEST IS PERFORMED BETWEEN 12 AND 20 WEEKS OF PREGNANCY.

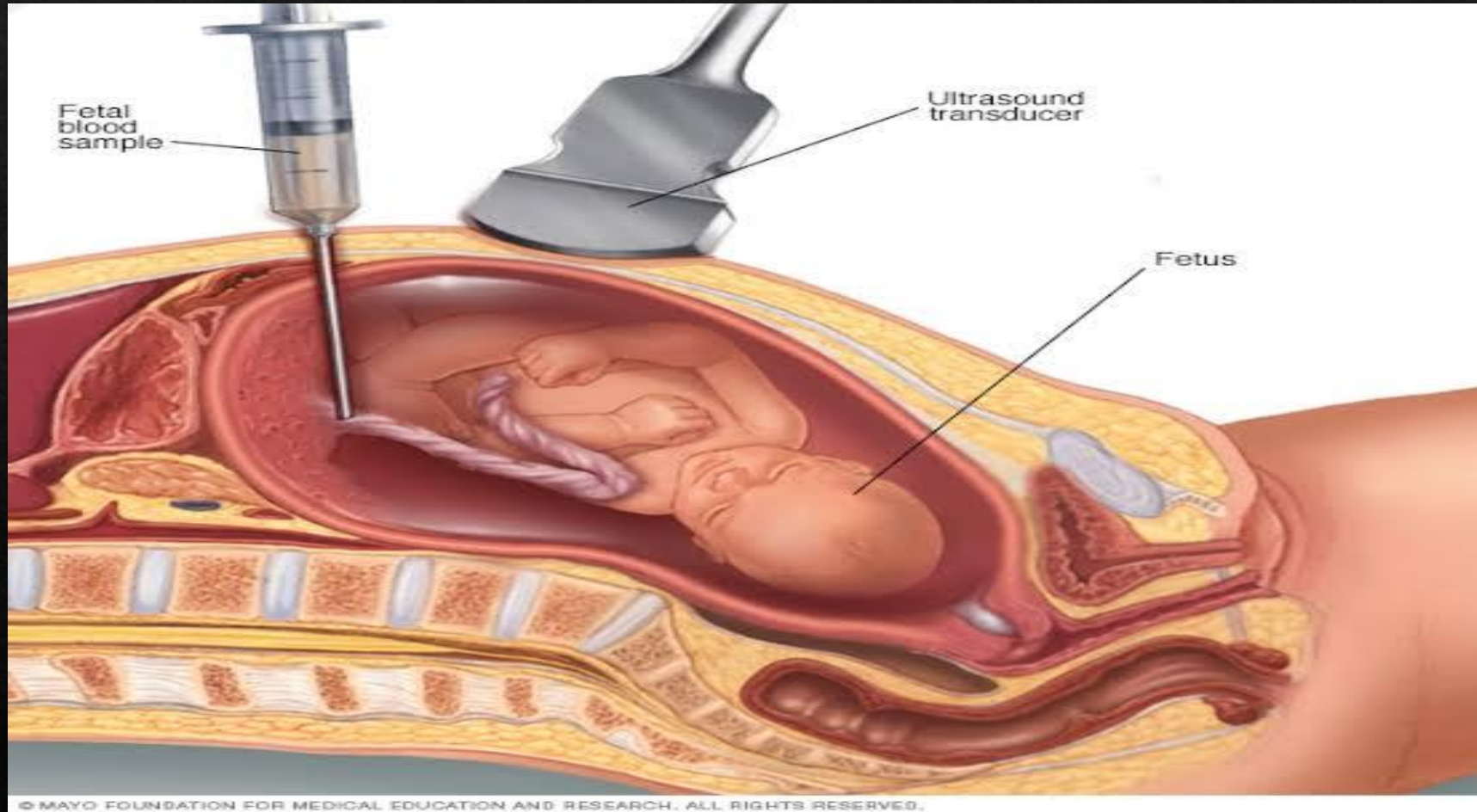
CHRONIC VILLUS SAMPLING & AMNIOCENTESIS



Nuchal translucency (NT) is investigated non-invasively through ultrasonography, while amniocentesis is invasive



PERCUTANEOUS UMBILICAL BLOOD SAMPLING



TREATMENTS

- ◆ “DOWN SYNDROME” is not a condition that can be cured.
- ◆ Treatment is directed at addressing the individual concerns of a particular individual (eg, certain heart defects may require surgery).
- ◆ Timely surgeries for cardiac and GI anomalies are necessary to prevent serious complications.
- ◆ Because the risk of vision problems , hearing loss, and infection is increased, screening and treatment may be necessary.

PREVENTION

- ◆ No prevention for down syndrome.
- ◆ Absolutely nothing that anyone can do to prevent a trisomy & there is nothing that anyone can do to cause trisomy.
- ◆ Fit out of prevention are aimed at genetic counseling of couples who are preparing to have babies screening test of AFP to determine chances-
- ◆ Teach parents the importance of food & fluids to maintain adequate nutrition.
- ◆ Emphasize the need to adequate nutrition. Poor feeding can result in obesity later in life.

**PREVENTION
WORKS!**

EDUCATION

- ◆ Teach family how to prevent physical complications.
- ◆ Avoid infection by engaging in good Handwashing.
- ◆ Increase fiber in diet to avoid constipation.
- ◆ Encourage physical activity.
- ◆ Advise parents to seek regular checkups for their child.

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Questions are the path to learning

Q₁] WHAT ARE THE
COMPLICATIONS OF
DOWN SYNDROME???????

Q₂] WHAT ARE THE
DIAGONSTIC TEST FOR
DOWN SYNDROME????



Thank
You