

DOWN  
SYNDROME  
[MEDICAL  
BIOLOGY]

BY:- SHUBHANSHRI  
JADHAV.



# 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

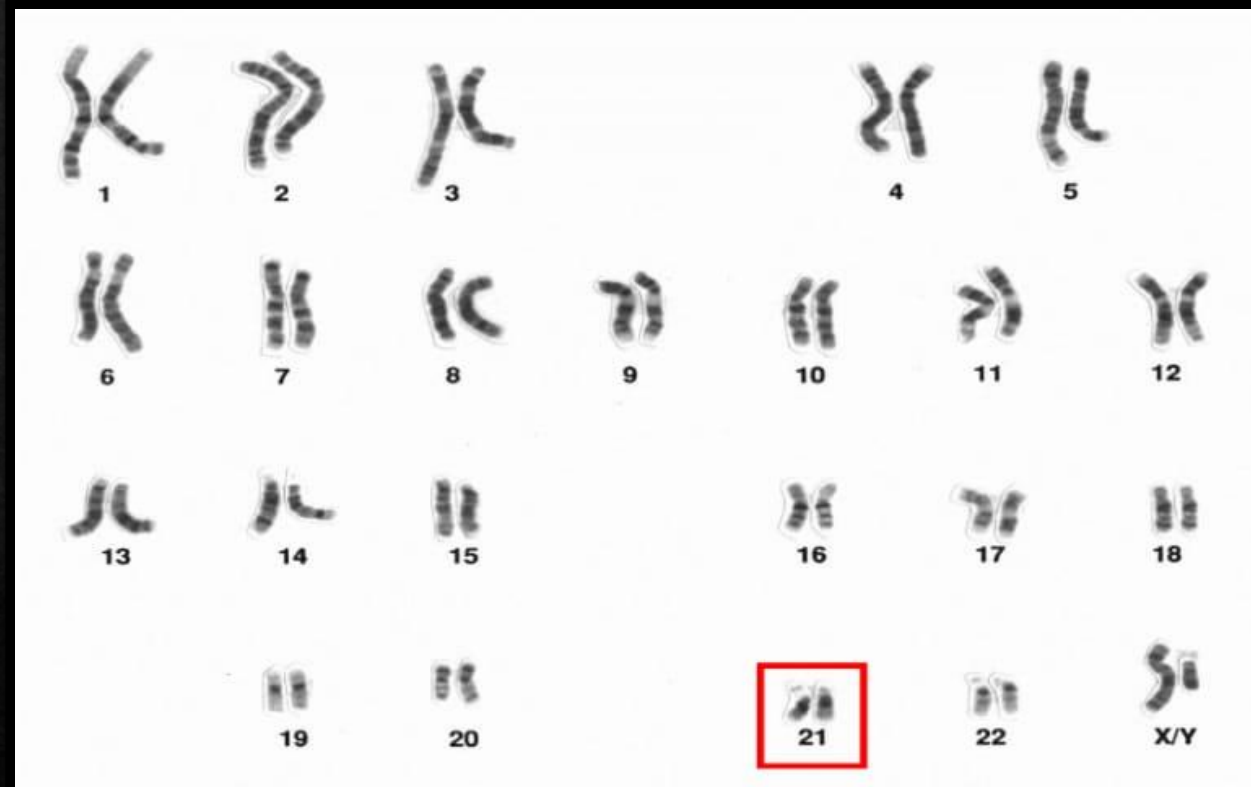
## TRANSLOCATIO N

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

## MOSIACISM

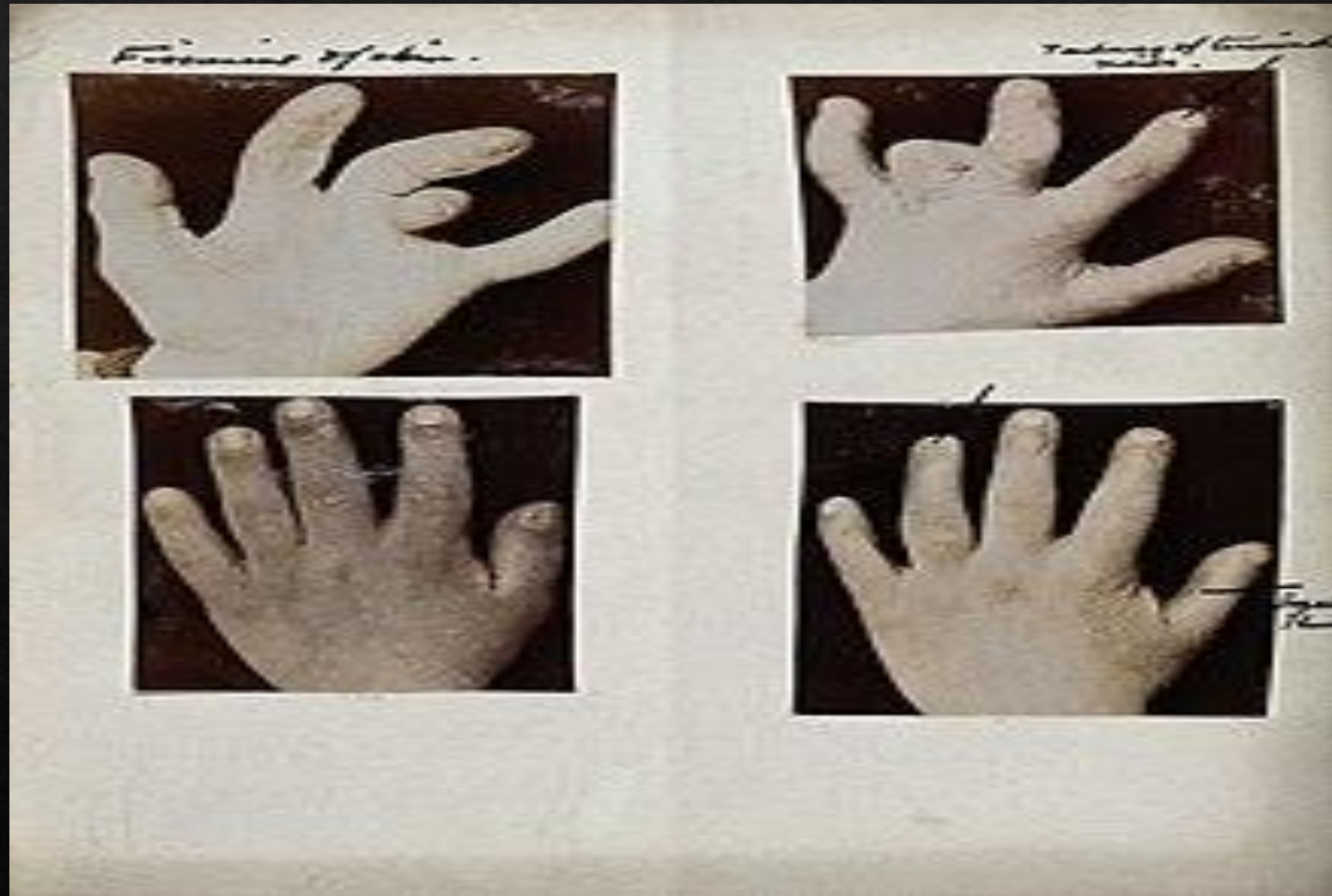
NONDISJUNCTION OCCURS AT A LARGER STAGE OF CELL DIVISION. THEREFORE, SOME CELLS HAVE THE NORMAL COMPLEMENT OF  $46$  CHROMOSOMES & OTHER CELLS  $47$  CHROMOSOMES.

# KARYOTYPE OF DOWN SYNDROME

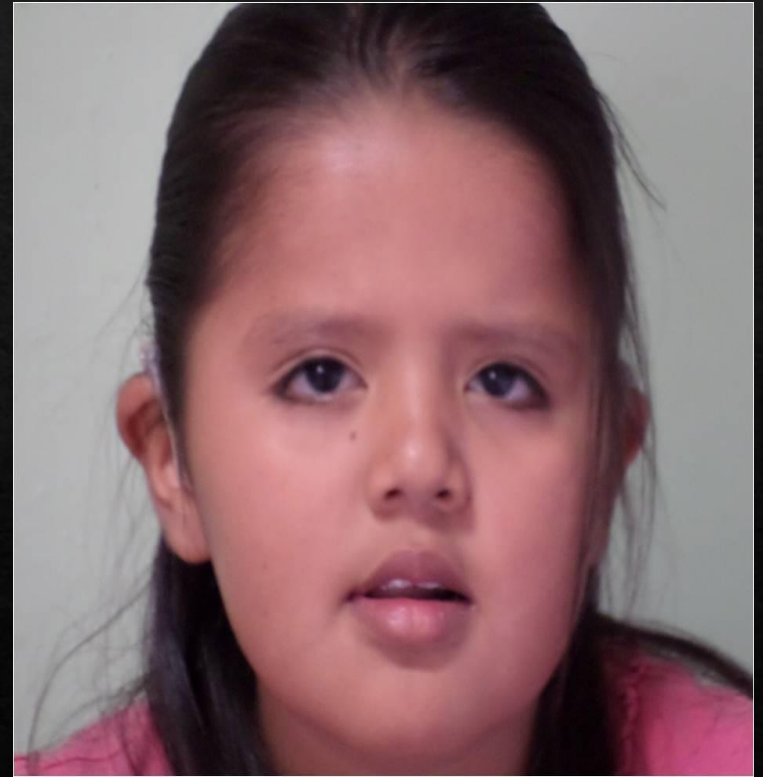


CLINICAL  
MANIFESTATIO  
N

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

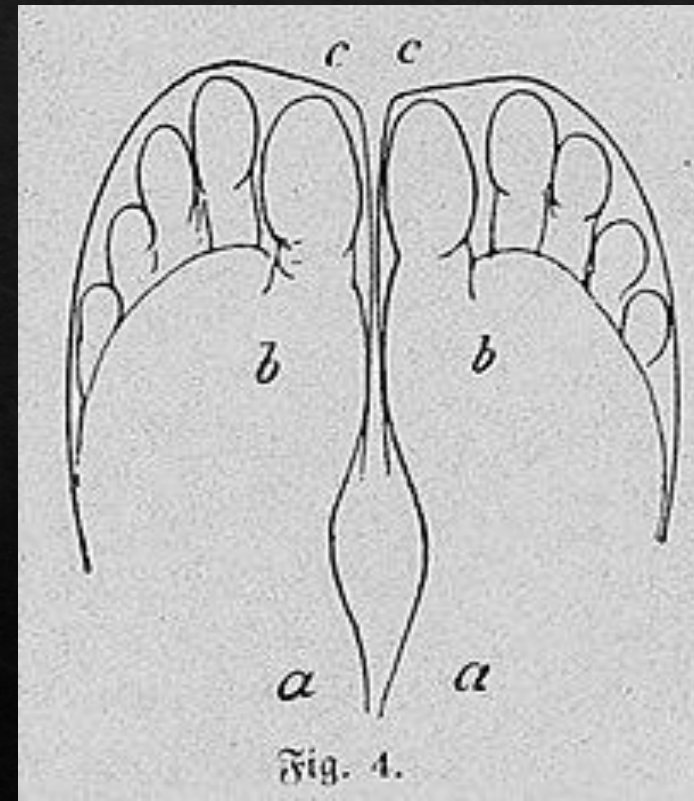


Fig. 4.

# 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.

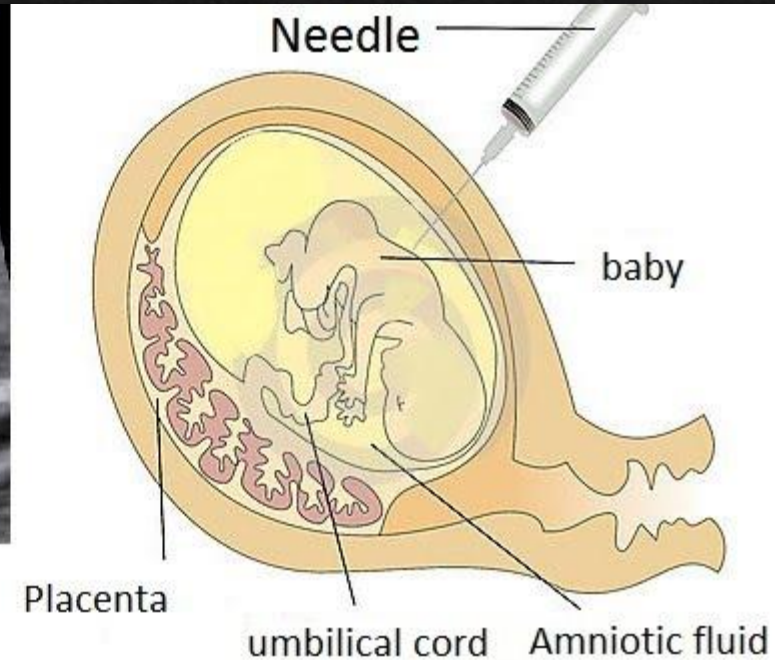
## 2] AMNIOCENTESIS

- ◆ 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.

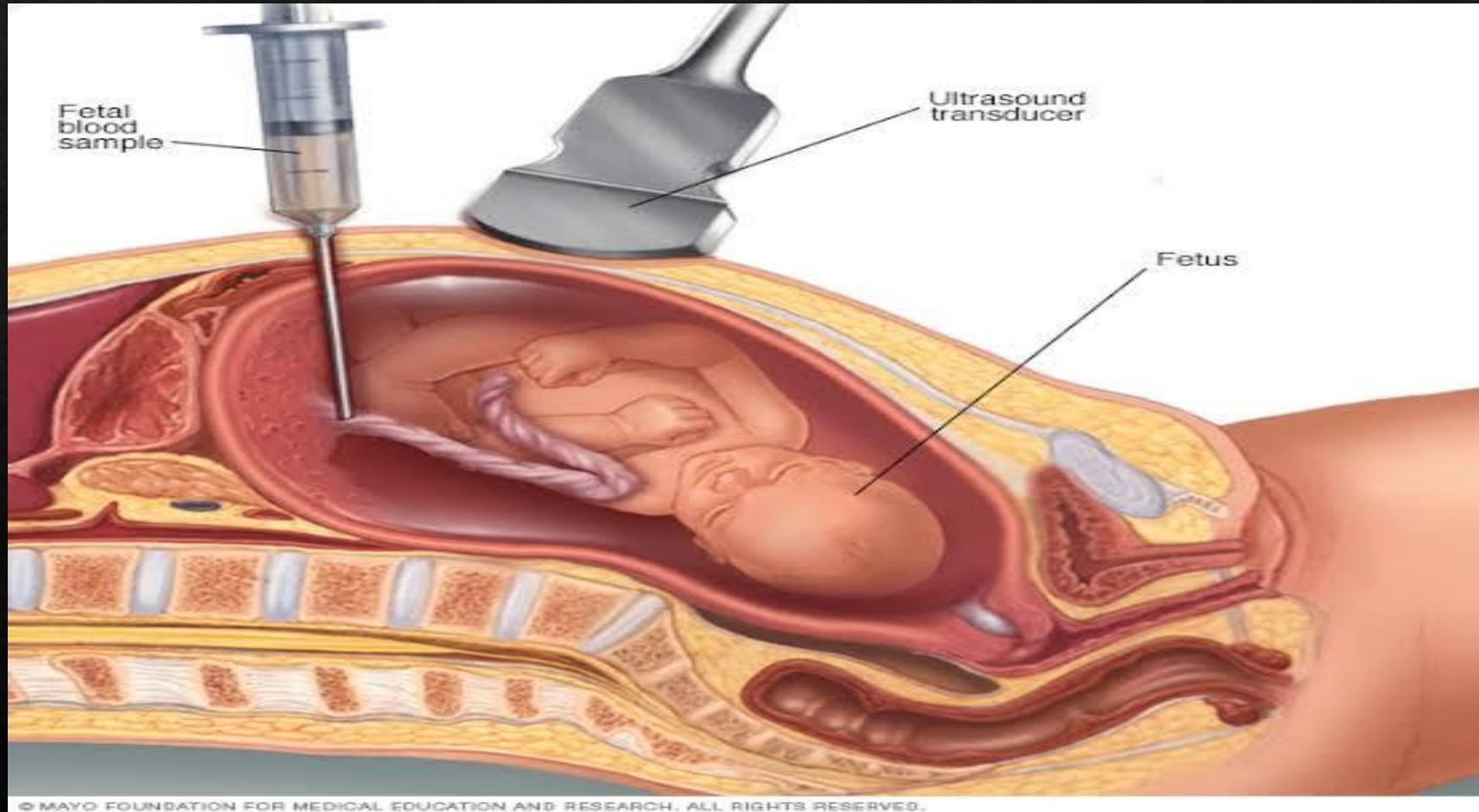
# CHRONIC VILLUS SAMPLING & AMNIOCENTESIS



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*

Q<sub>1</sub>] WHAT ARE THE  
COMPLICATIONS OF  
DOWN SYNDROME???????

Q<sub>2</sub>] WHAT ARE THE  
DIAGONSTIC TEST FOR  
DOWN SYNDROME????



Thank  
You