

# *Down Syndrome*

**Dr. Langdon Down**

Who described it first **in 1866**

# SYNDROME

- Group of symptoms which consistently occur together
- Combination of opinions, emotions or behaviour that is typical of particular group of people

# Genotype

- **95%- 47,XX or 47,XY**
- **4%- Translocation  $\frac{21}{14}$  46,XX or XY  
14,+t ( 14q 21q)**
- **1%- Mosaic- 47,XX/46,XX or 47,XY/46,XY**

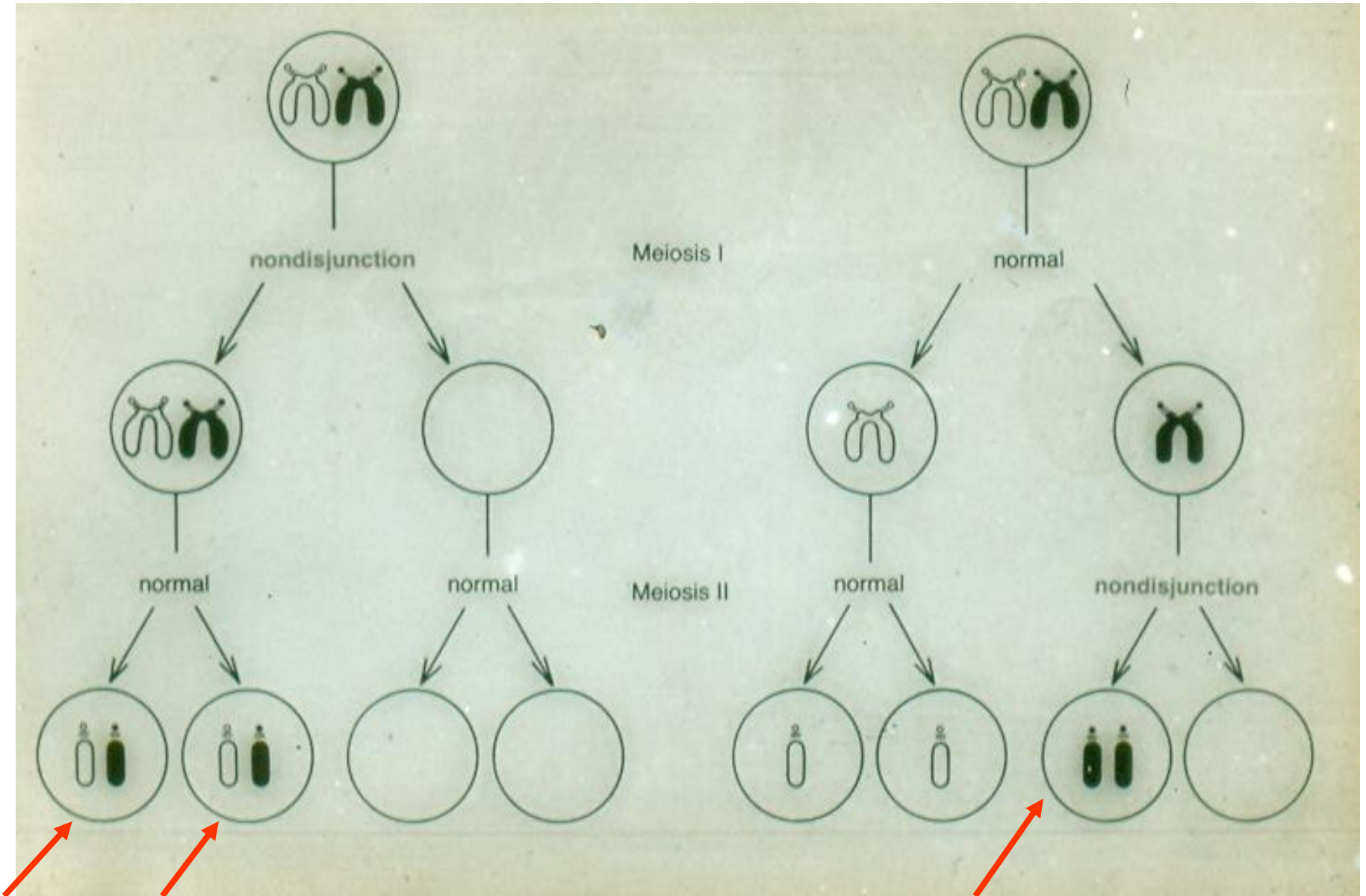
# Causative factors

- Late Maternal age-by far most common
- Inherited from the Mother with Translocation

# Cytogenetics

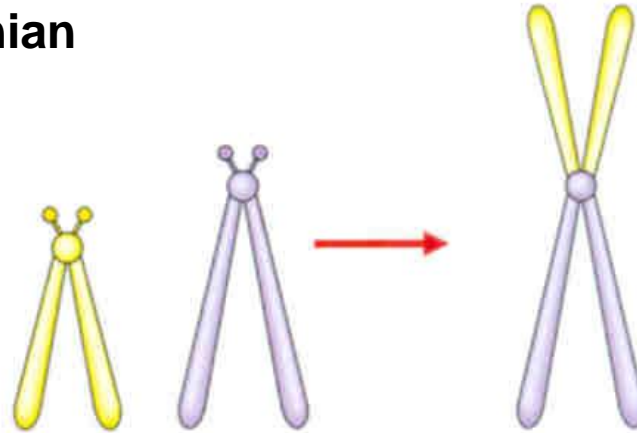
- **NONDISJUNCTION** at **M-I** or **M-II-**  
**47,XX** or **47, XY**
- Inherited from Mother who is  
**TRANSLOCATION CARRIER**
- **MOSAIC-** **Post-zygotic NONDISJUNCTION**  
Genotype may vary in the same person as age advances

# NONDISJUNCTION



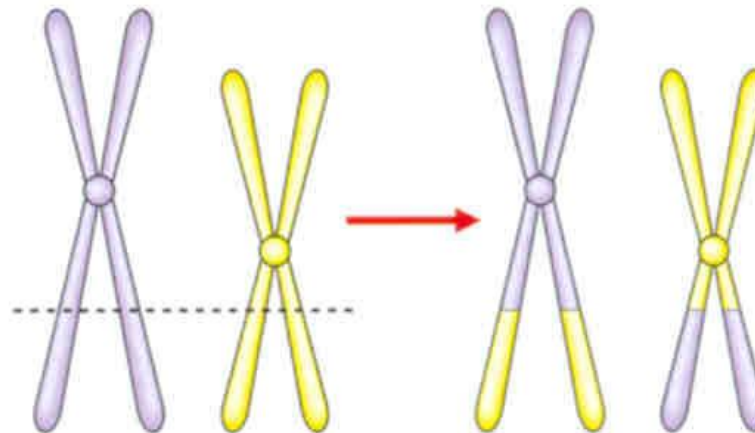
# TRANSLOCATION

**Robertsonian**



**Unbalanced**

**Reciprocal**



**Balanced**

21



14



21  

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14

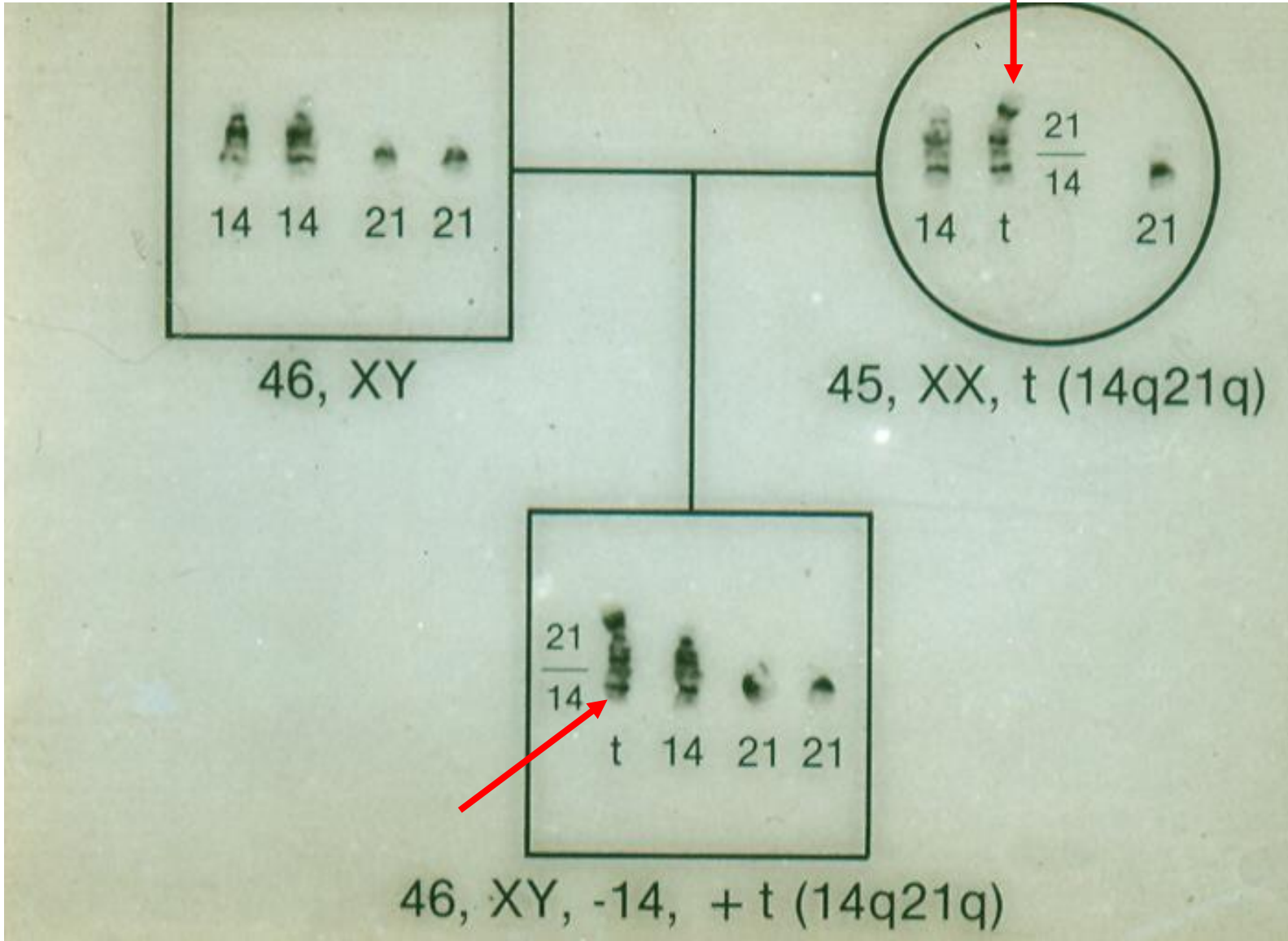
Translocation



Robertsonian  
translocation



# Effects of Translocation carrier mother

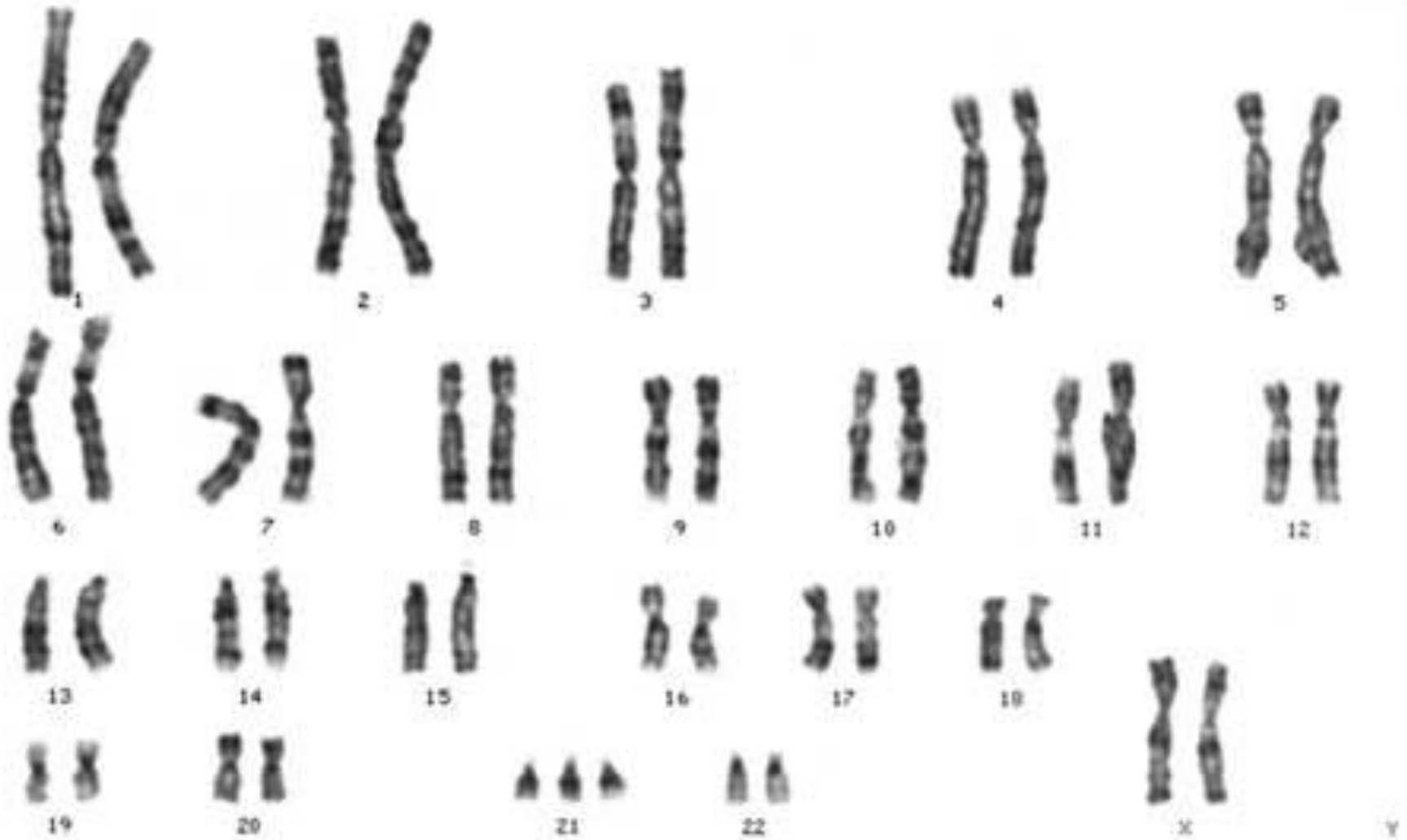


- Concordance in monozygotic twins
- Discordance in dizygotic twins

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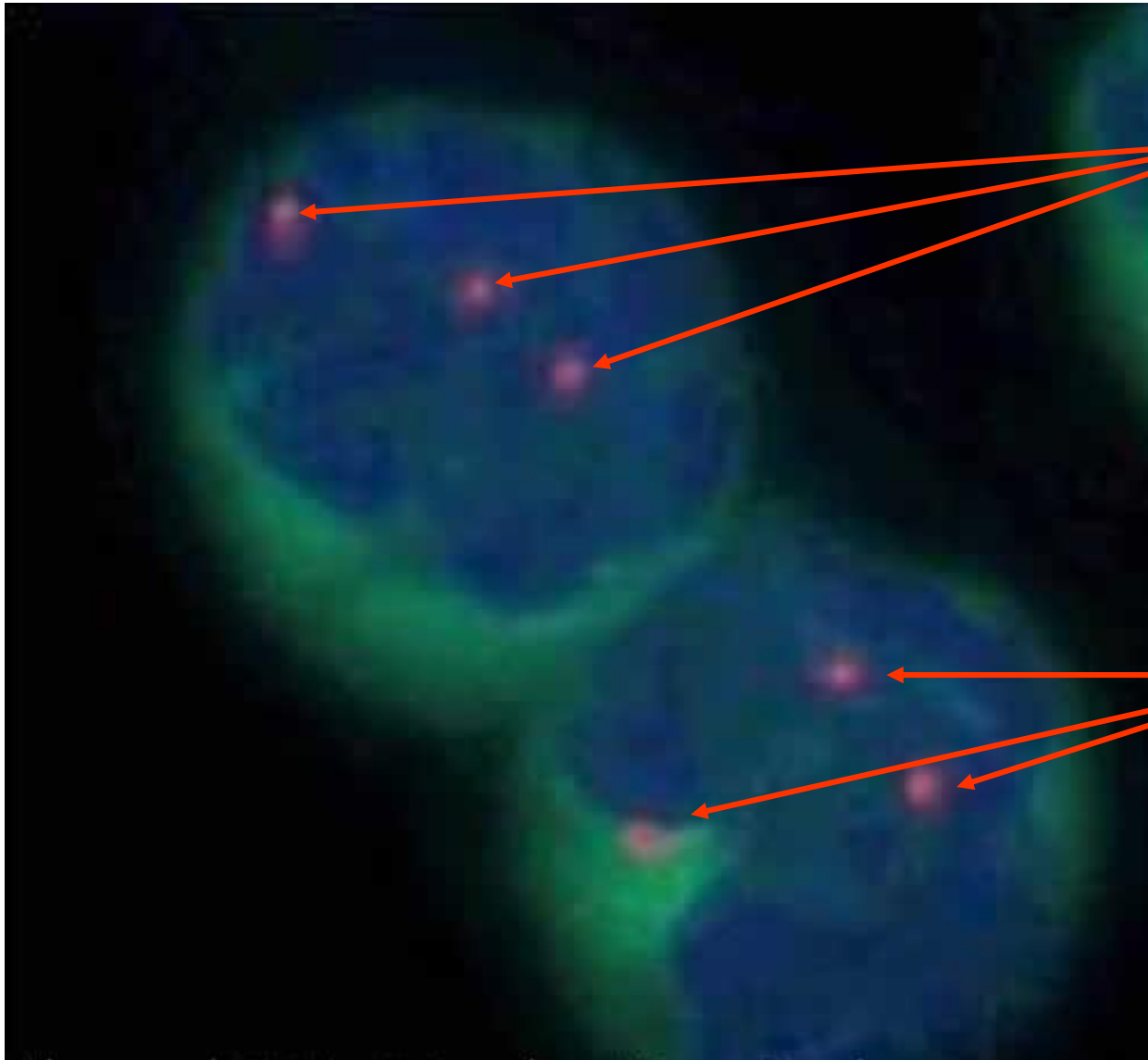
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# FISH for chromosome 21



# FISH For chromosome 21



# Phenotype

## Sex

**Since it is Autosomal Aberration**

**Males and females are equally affected**

**SEX NO BAR**

**3/4<sup>th</sup> of the patients-**

**spontaneous abortions**

**Many die in early infancy due to -**

**cardio-pulmonary insufficiencies**

# INCIDENCE

**1:800 to 1:1000**

Strong Association between the  
Incidence  
and

Advancing Maternal Age

**1:400(35yrs) to 1:100(40years)**



# Main features

- **Neuromuscular disorder**
- **Low I Q- Mentally Retarded**

# IQ

## LOW

- 25 to 75
- Average – 40 to 45

# At Birth Severe Hypotonia



Observe- Hypotonia  
Protruding tongue  
Child lies floppy on the bed



# Down Syndrome



# Signs and symptoms

## AT BIRTH

- Hypotonia
- Sleepy
- Poor sucking reflex-  
First noticed by the mother
- Lack of Morrow's reflex

# Prompt Rapid Diagnosis

- Hypotonia
- Characteristic upward slant of palpebral fissure
- Small ears
- Protruding tongue

# EARLY CHILDHOOD

Since it is NEUROMUSCULAR disorder

- Delayed milestones-  
like Turning, Crawling, Sitting, Walking
- Low I Q



# Craniofacial

- Flat Occiput - Brachycephaly
- Protruding tongue
- Small ears
- Upward slant of Palpebral fissure

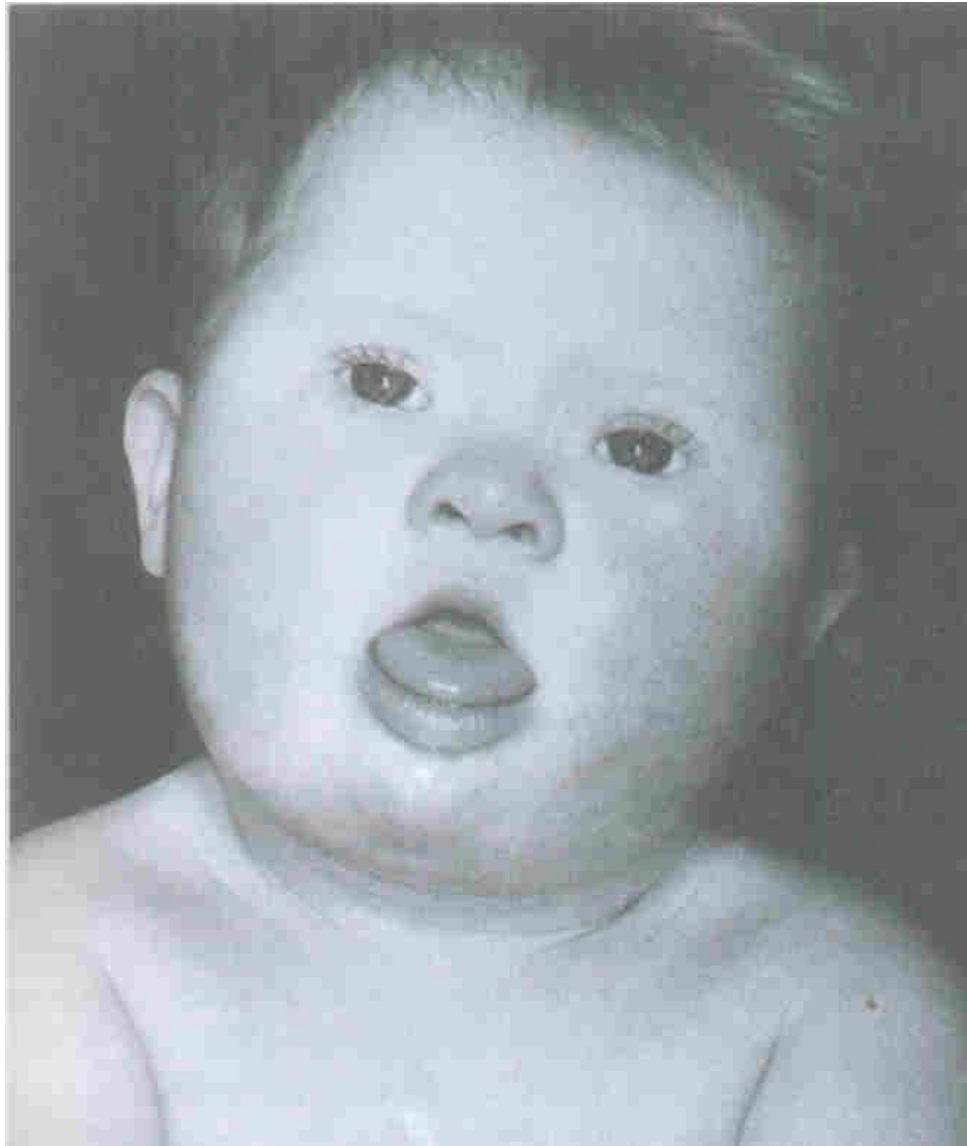
# Craniofacial -contd

- Brushfield Spot in the iris
- Squinting either in the childhood or in later years
- Low bridge nose

Upward slant, Epicanthic folds, Brushfield spots, Low bridged Nose



# Protruding Tongue



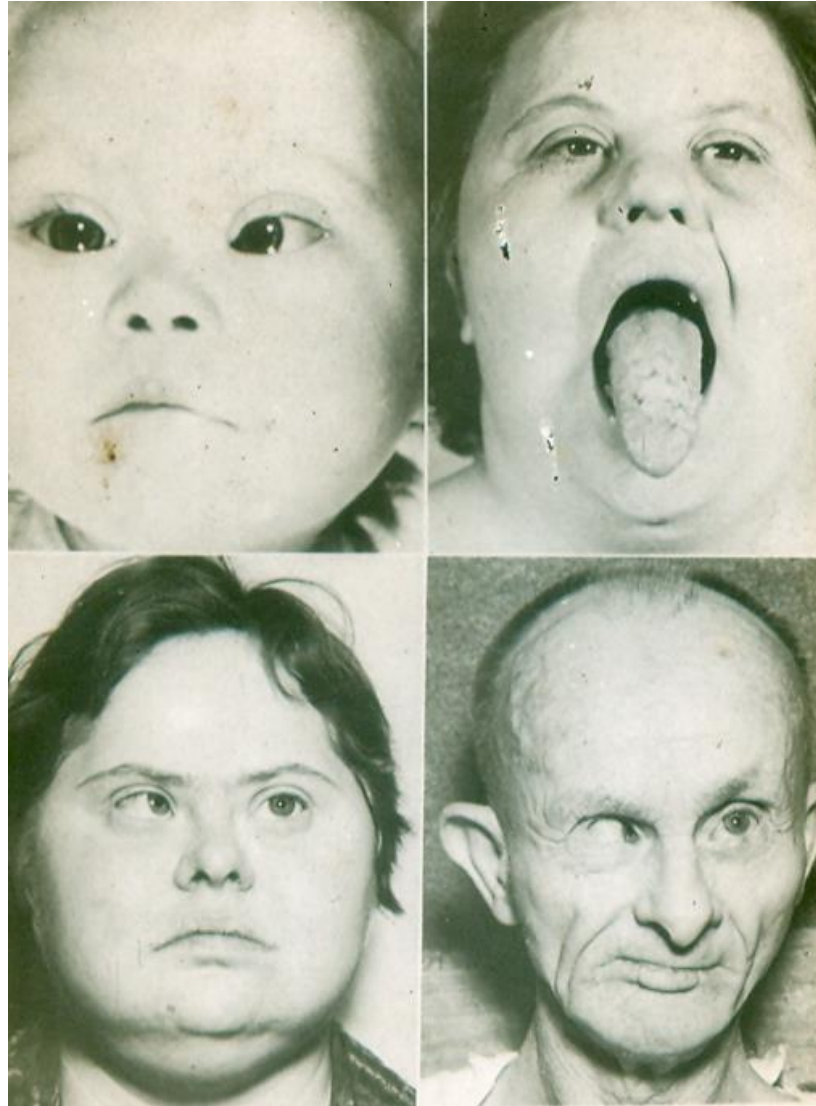
# Squint



# Brushfield spots



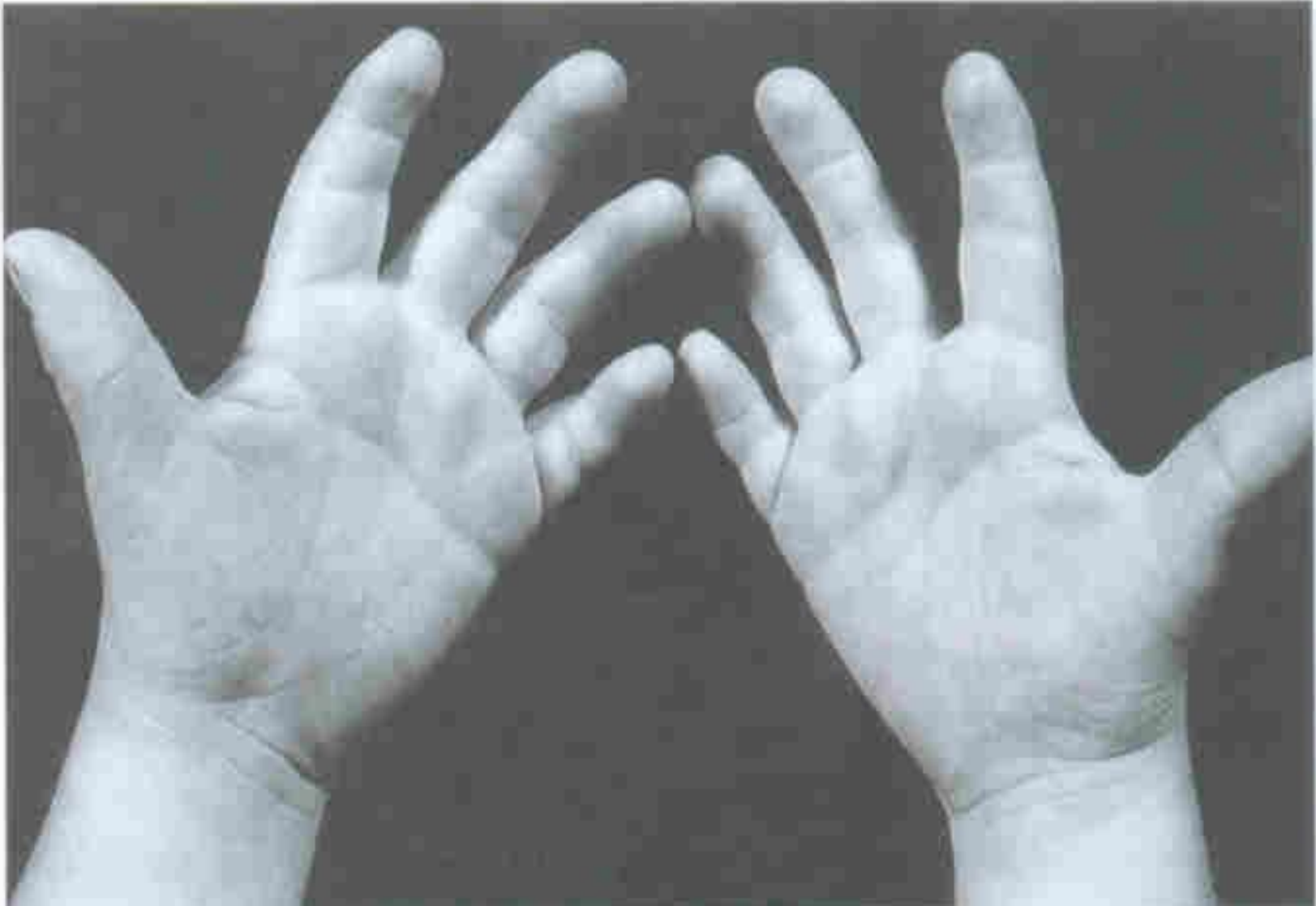
# Fissured tongue, Squinting in babies and in old age



# Limbs

- Single Palmar crease
- Clinodactyly-Small and inturned little finger
- Wide gap between first and second toe

# Simian crease, Clinodactyly





# Single Palmer or SIMIAN Crease

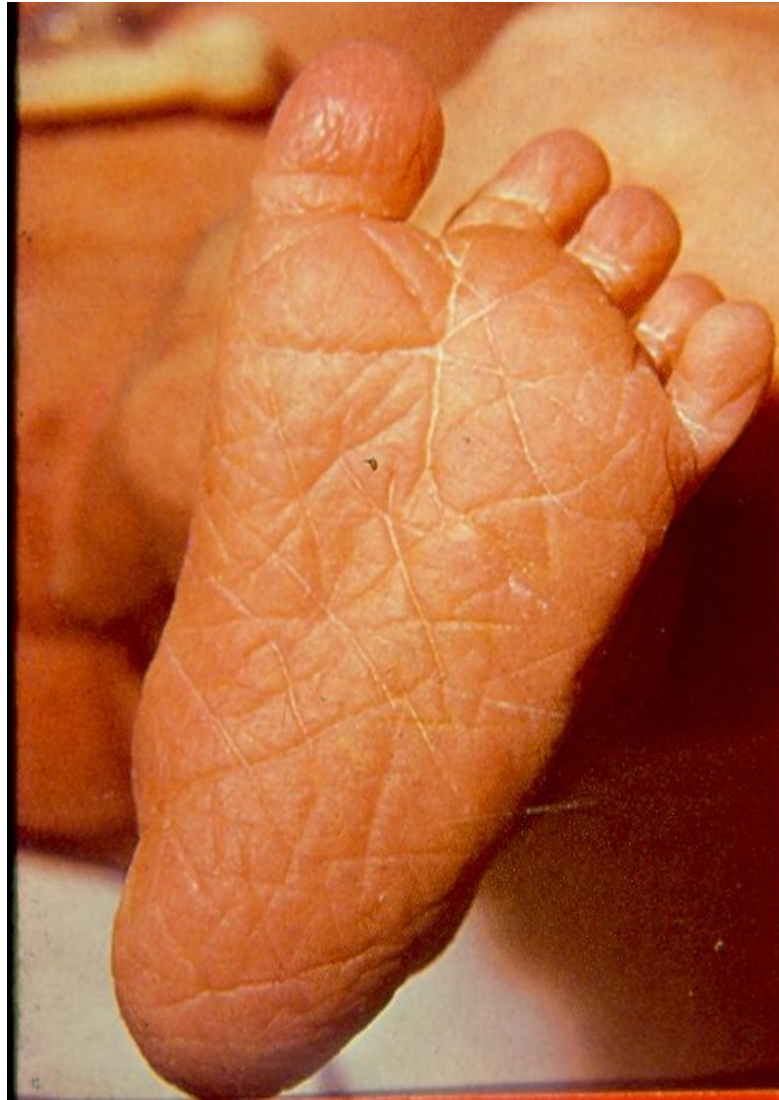




# Simian Crease 9-10 ulnar loops



# Gap between First and Second toe



# Reproductive System

- Females are **FERTILE**
- Males are **STERILE**

# Males Sterile, Females- Fertile Mother with Down with Normal Baby



# Behavioral Details

# Happy and Affectionate





# Sense of Rhythm, Love to Dance



# Love music, Faculty of mimicry, but Adamant



Educable and can be of help





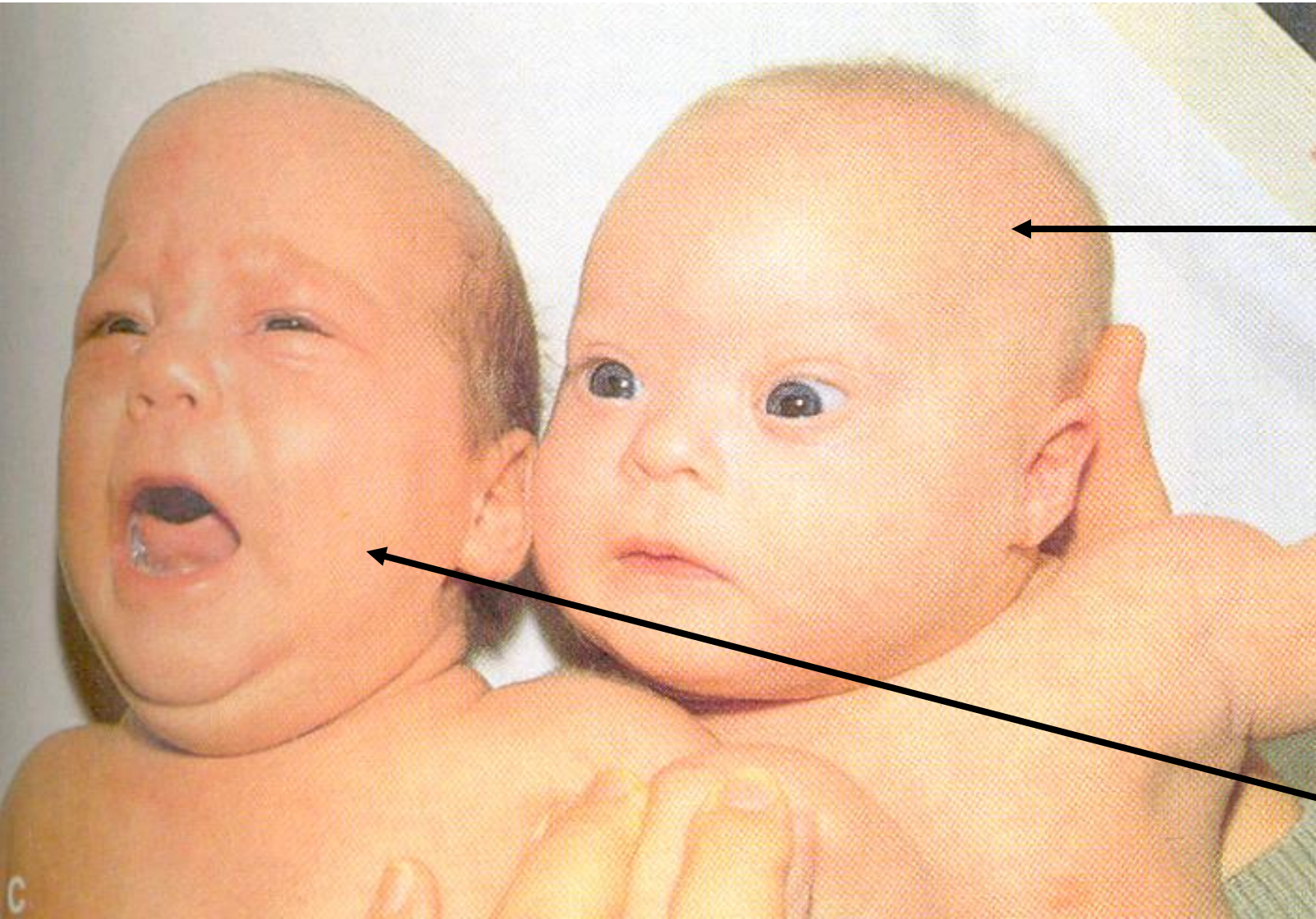
Can lead  
Semi-normal life

Observe Squinting

# Average Age-50 years

- 15 times more prone to **Leukemia**
- Most Adults develop  
**Alzheimer Disease**

# Discordant in Dizygotic twins



21 Trisomy

Normal

# Dizygotic twins

Baby on the Right has 21TRISOMY

Twin sister on the Left is NORMAL



# Mosaic- Improved appearance at later stage





# Down Syndrome- Mosaic

Note the change in the appearance



# Counseling

- Pre-marriage
- Pre-pregnancy
- Antenatal
- Post-natal

# Antenatal

- Maternal alpha-feto-proteins– **LOW**
- Karyotyping through Amniocentesis with advanced maternal age
- Karyotyping of the unusually dull mother to rule out Translocation carrier

1. Following is the genotype of Down syndrome

a) 47,XX or 47,XY

b) 45,XX or 45,XY

c) 47XXX or 47,XXY

d) 45 XO

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## 2. Following is/are the causes of TRISOMY 21

- a) Non disjunction at Meiosis
- b) Offspring of Translocation mother
- c) Mosaicism-Post-zygotic non-disjunction-
- d) All of the above

## 2. Following is/are the causes of TRISOMY 21

- a) Non disjunction at Meiosis
- b) Offspring of Translocation mother
- c) Post-zygotic non-disjunction-Mosaicism
- d) All of the above**

### 3. Following statements true regarding Down Syndrome

a) It is Trisomy 21

b) It is Neuro-Muscular disorder

c) Females are sterile

d) Hypotonia is the first sign noticed



### 3. Following statements true regarding Down Syndrome

a) It is trisomy 21

b) It is Neuro-Muscular disorder

**c) Female are sterile**

d) Hypotonia is the first sign noticed



[catpalsale.com](http://catpalsale.com)

**THANK YOU**

It is deep, wrenching feeling  
in your stomach,

when

the pages of your text book still smell new

and

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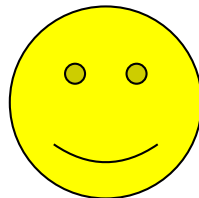
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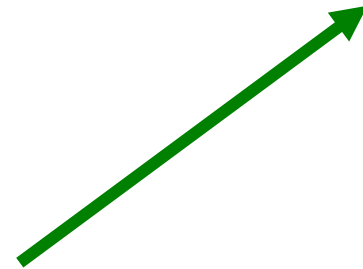
Just few hours are left for your **EXAM.**



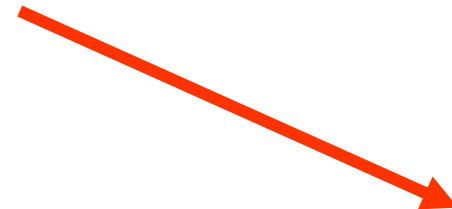
# BEST OF LUCK

DO BETTER THAN THE PREVIOUS EXAM

GRAPH SHOULD BE



And not like

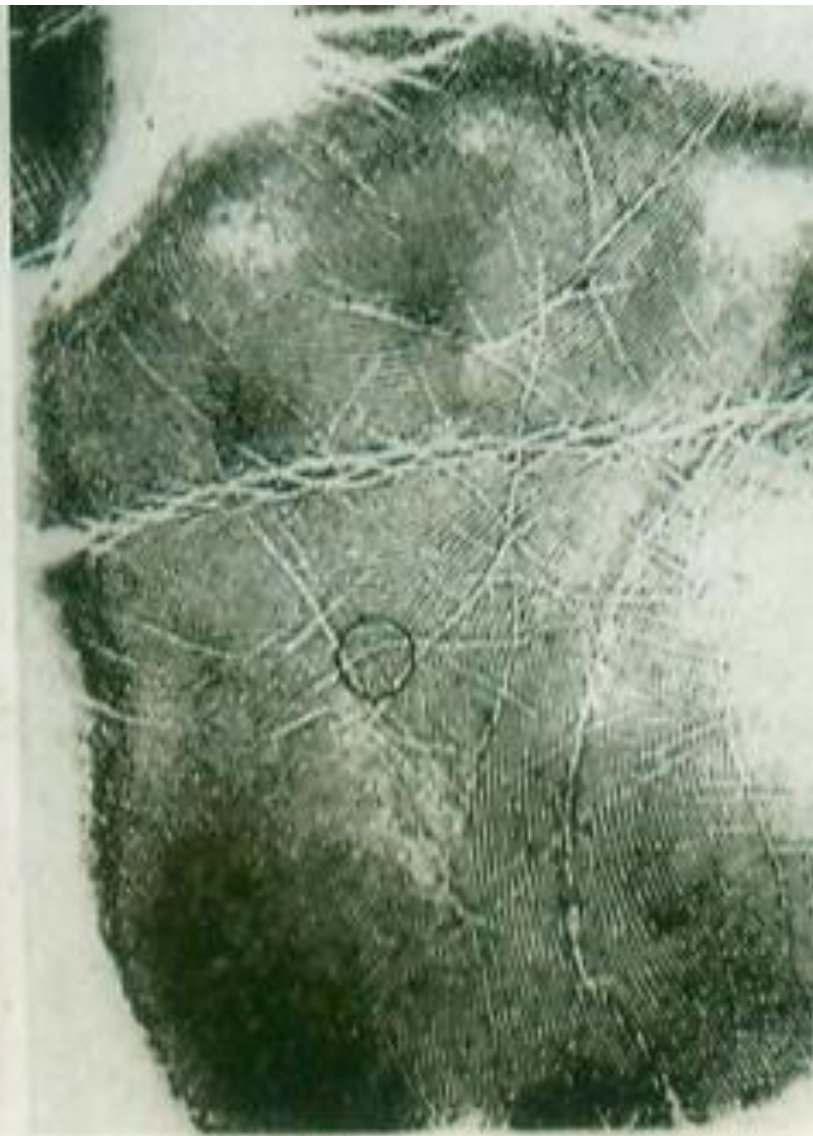












# Epicanthic fold and Brushfield spots

