

**IMPACT AND IMPLICATIONS**

**OF**

**CHROMOSOMAL**

**ABNORMALITIES**

- **CHROMOSOMES:**
- **IMBALANCE CAUSED BY-**
- **(A) ABSENCE**
- **(B) DUPLICATION OF MATERIAL**

- **TWO TYPES OF CHROMOSOMAL**
- **IMBALANCE:**
- **1.NUMERICAL ABNORMALITIES**
- **(Aberations in Chromosomes**
- **Number)**
- **2.DEFECT IN STRUCTURE:**
- **DIAGNOSIS BY KARYOTYPING**

# NUMERICAL ABERRATIONS

- **1. TRISOMY: ASSOCIATED WITH**
- **ADVANCED MATERNAL AGE**
  
- **2. MONOSOMY: SEGREGATION**
- **ERROR DURING CELL DIVISION.**

# NUMERICAL ABERRATIONS

- **3.POLYPOID: DUE TO EXTRA SET**
- **OF CHROMOSOMES.**
- **4.TRIPLOIDY: TWO SPERMATOZOA**
- **FERTILIZE ONE**
- **OOCTE---ZYGOTE—CONTAINS**
- **THREE SETS OF CHROMOSOMES**
- **INSTEAD OF TWO SETS.**

# NUMERICAL ABERRATIONS

**DONOT REPEAT IN SUBSEQUENT  
PREGNANCIES.**

**PARENTAL CHROMOSOMES  
ARE NORMAL.**

# STRUCTURAL ANOMALIES

- **DEFECT IN STRUCTURE OF**
- **ONE CHROMOSOME:**
  - **1. INVERSION: TURNED AROUND**
  - **2. RINGS**
  - **3. TRANSLOCATION**

# STRUCTURAL ANOMALIES

- **TRANSLOCATION:**
- **RECEPROCAL**
- **ROBERTSONIAN**
- **ACCROCENTRIC CHROMOSOMES**
- **ARE 13,14,15,24, 21&22.**



- **STRUTURAL ANOMALIES**
- **FOUND IN FOETUS**
- **PARENTS KARYPTING IS**
- **NECESSARY**

# CONGENITAL ANOMALY

- RECEPROCAL TRANSLOCATION IN
- WHICH A SEGMENT OF CHROMOSOME EXCHANGED WITH
- A SEGMENT OF NONHOMOLOGOUS
- CHROMOSOME LEADING TO GAMETE FORMATION WITH
- UNBALANCED AMOUNT OF
- CHROMOSOMAL MATERIAL .

# THROMBOCYTOPENIA

- **IMPAIRED PLATELET DESTRUCTION**

**DECREASED PLATELET PRODUCTION**

**ABNORMAL POOLING OF PLATELETS**

# THROMBOCYTOPENIA

- **INCREASED PLATELET DESTRUCTION**
- 
- **IDIOPATHIC --- SECONDARY**
- **SECONDARY = IMMUNE TYPE**
- **NON IMMUNE TYPE**

# SECONDARY THROMBOCYTOPENIA

- IMMUNE MECHANISM=
- COLLAGEN VASCULAR DISEASES
- LYMPHO PROLIFERATIVE DISORDERS
- S.L.E, PREGNANCY
- DRUGS=ACETAZOLAMIDE,DIGOXIN,
- GOLD SALTS,HEPARIN,QUINIDINE,
- ACETAMINOPHEN

# **NON IMMUNE MECHANISM**

- **D.I.C, I.T.P, APLASTIC ANAEMIA**
- **ACUTE LEUKAEMIA**
- **MYELOSUPPRESSIVE DRUGS**
- **CYCLOPHOSPHAMIDE**
- **BUSALPHAN**

- **CIRRHOSIS OF LIVER**
- **FOLATE DEFICIENCY**
- **VIT B12 DEFICIENCY**
- **HYPERTENSION**
- **I.T.P(idiopathic thrombocytopenic  
purpura)**
- **T.T.P(thrombotic-----do-----)**
- **H.U.S(Hemolytic uraemic syndrome)**

# MEDICAL TERMINATION OF PREGNANCY(M.T.P)

- **CONSANGUINITY**
- **MEPHEPRISTINE (RO 486)**
- **DAY -1 --- 3tabs**
- **MESOPROSTAL**
- **DAY-3 ---- 4 tabs**