

BACKGROUND

- **ABNORMAL NUMBER of CHROMOSOMES in a CELL**
 - ~ COMMON CAUSE of GENETIC DISORDERS

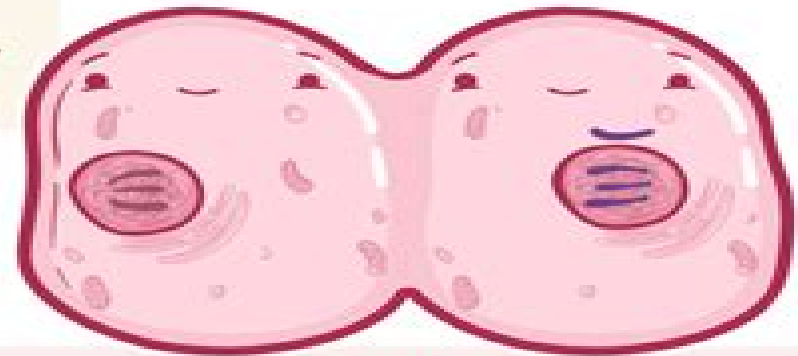
SIGNS & SYMPTOMS

- **MOST ANEUPLOIDIES INCOMPATIBLE with LIFE & RESULT in SPONTANEOUS ABORTIONS in FIRST WEEKS of PREGNANCY**
- **3 VIABLE AUTOSOMAL ANEUPLOIDIES:**
 - ~ TRISOMY 21 (DOWN SYNDROME)
 - ~ TRISOMY 13 (EDWARDS SYNDROME)
 - ~ TRISOMY 18 (PATAU SYNDROME)
- **COMMON SEX CHROMOSOME DISORDERS:**
 - ~ KLINEFELTER SYNDROME
 - ~ TURNER SYNDROME



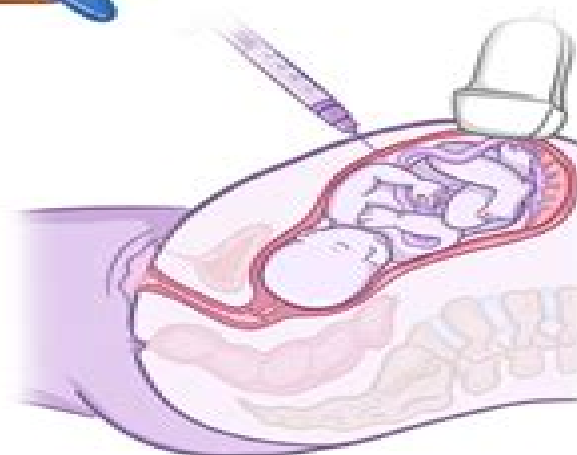
DIAGNOSIS

- **KARYOTYPE ANALYSIS**
 - ~ PRENATALLY via CHORIONIC-VILLUS SAMPLING between 9-11 WKS
 - ~ AMNIOCENTESIS between 15-20 WKS
- **PRENATAL SCREENING**
 - ~ COMBINED TEST DURING 1st TRIMESTER: of NUCHAL TRANSLUCENCY SCAN, MEASURING PAPP-A & beta-hCG SERUM HORMONE LEVELS
 - ~ 2nd TRIMESTER: QUADRUPEL TEST MEASURING AFP SERUM LEVELS, uE3, beta-hCG, & INHIBIN A
 - ~ ALTERNATIVE: ASSESSMENT of CELL-FREE DNA in MATERNAL BLOOD SAMPLE, ANALYZING PRESENCE of FETAL DNA in MATERNAL BLOOD (EARLY as 10 WKS)



CAUSES

- **NONDISJUNCTION during CELL DIVISION**
 - ~ MEIOSIS I: ONE CHROMOSOME PAIR DOESN'T SPLIT
 - ~ MEIOSIS II: SISTER CHROMATID DOESN'T SEPARATE
- **ADVANCED MATERNAL AGE ↑↑ RISK**
 - ~ > 20 YO: 1/1500 BIRTHS
 - ~ > 45 YO: 1/25 BIRTHS
- **ROBERTSONIAN TRANSLOCATION**
- **WHEN ANEUPLOIDIES NOT PRESENT in ALL CELLS: MOSAICISM**



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