

BACKGROUND

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

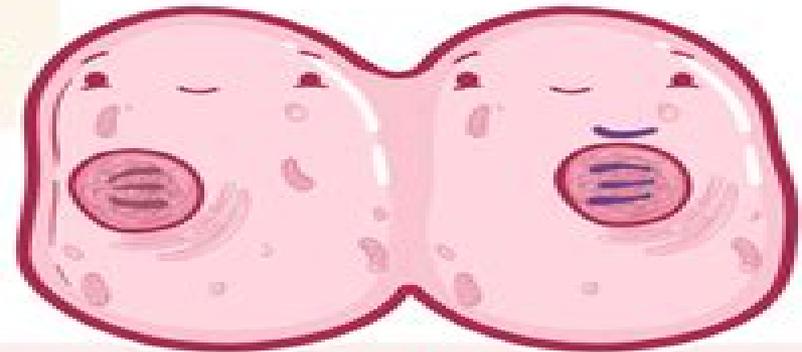
SIGNS & SYMPTOMS

- ≠ **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**



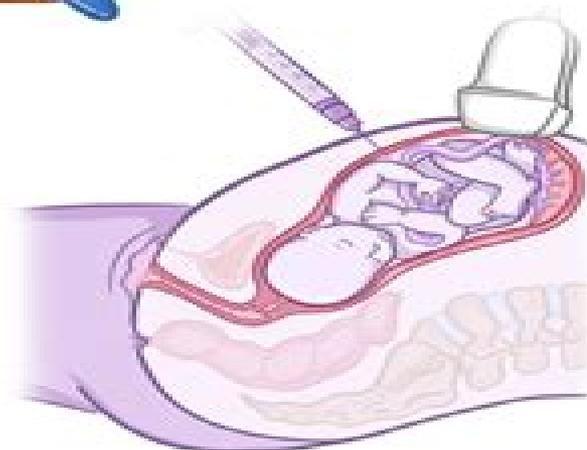
DIAGNOSIS

- ≠ **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: QUADRUPLE TEST** **MEASURING AFP SERUM LEVELS, uES, 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**



Thalabaemia Syndromes

Anthony S. Fauci



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Table of Contents Thalabaemia Syndromes

1. Understanding the eBook Thalabaemia Syndromes
 - The Rise of Digital Reading Thalabaemia Syndromes
 - Advantages of eBooks Over Traditional Books
2. Identifying Thalabaemia Syndromes
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Thalabaemia Syndromes
 - User-Friendly Interface
4. Exploring eBook Recommendations from Thalabaemia Syndromes
 - Personalized Recommendations
 - Thalabaemia Syndromes User Reviews and Ratings
 - Thalabaemia Syndromes and Bestseller Lists
5. Accessing Thalabaemia Syndromes Free and Paid eBooks
 - Thalabaemia Syndromes Public Domain eBooks
 - Thalabaemia Syndromes eBook Subscription Services

- Thalabaemia Syndromes Budget-Friendly Options
- 6. Navigating Thalabaemia Syndromes eBook Formats
 - ePub, PDF, MOBI, and More
 - Thalabaemia Syndromes Compatibility with Devices
 - Thalabaemia Syndromes Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Thalabaemia Syndromes
 - Highlighting and Note-Taking Thalabaemia Syndromes
 - Interactive Elements Thalabaemia Syndromes
- 8. Staying Engaged with Thalabaemia Syndromes
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Thalabaemia Syndromes
- 9. Balancing eBooks and Physical Books Thalabaemia Syndromes
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Thalabaemia Syndromes
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Thalabaemia Syndromes
 - Setting Reading Goals Thalabaemia Syndromes
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Thalabaemia Syndromes
 - Fact-Checking eBook Content of Thalabaemia Syndromes
 - Distinguishing Credible Sources
- 13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
- 14. Embracing eBook Trends

- Integration of Multimedia Elements
- Interactive and Gamified eBooks

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