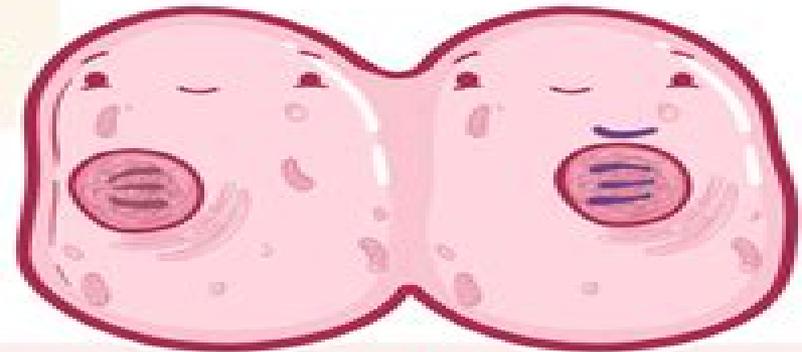


## BACKGROUND

- ≠ **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**

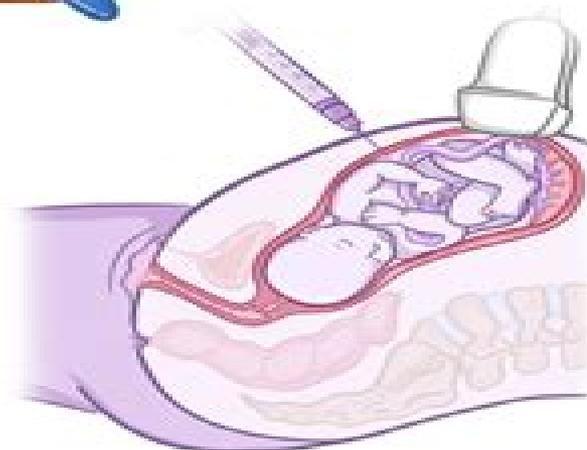


## CAUSES

- ≠ **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**

## 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)**



# Thalabaemia Syndromes

**JS Bruner**



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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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