

BACKGROUND

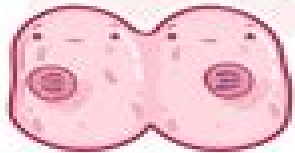
* **RARE GENETIC DISORDER CHARACTERIZED by COMPLEX CRANIOSYNOSTOSIS, BROAD THUMBS, & SKELETAL DEFECTS AFFECTING HANDS & FEET**

- AUTOSOMAL DOMINANT (ONLY 1 COPY of MUTATED GENE NEEDED)
- PREMATURE FUSION of BONES in SKULL, HANDS, & FEET



CAUSES

- * **MUTATION in FGFR1 & FGFR2 GENES**
 - AFFECT HOW CELLS GROW, DIVIDE, & DIE

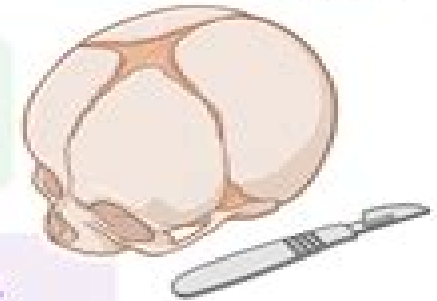


DIAGNOSIS

- * CLINICAL MANIFESTATIONS
- * GENETIC TESTING

TREATMENT

- * **RECONSTRUCTIVE SKULL SURGERY**



SIGNS & SYMPTOMS

- * WIDE & HIGH FOREHEAD
- * MIDFACE HYPOPLASIA
- * OCULAR HYPERTELORISM
- * SMALL NOSE with LOW BRIDGE
- * BROAD THUMBS & BIG TOES
- * SYNDACTYLY
- * HEARING LOSS
- * VISUAL DEFECTS
- * RESPIRATORY PROBLEMS
- * LEARNING DISABILITIES & DEVELOPMENTAL DELAYS (SPEECH & LANGUAGE)



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Table of Contents Syndrom D

1. Understanding the eBook Syndrom D
 - The Rise of Digital Reading Syndrom D
 - Advantages of eBooks Over Traditional Books
2. Identifying Syndrom D
 - 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 Syndrom D
 - User-Friendly Interface
4. Exploring eBook Recommendations from Syndrom D
 - Personalized Recommendations
 - Syndrom D User Reviews and Ratings
 - Syndrom D and Bestseller Lists
5. Accessing Syndrom D Free and Paid eBooks
 - Syndrom D Public Domain eBooks
 - Syndrom D eBook Subscription Services
 - Syndrom D Budget-Friendly Options
6. Navigating Syndrom D eBook Formats
 - ePub, PDF, MOBI, and More
 - Syndrom D Compatibility with Devices
 - Syndrom D Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Syndrom D
 - Highlighting and Note-Taking Syndrom D
 - Interactive Elements Syndrom D
8. Staying Engaged with Syndrom D

- Joining Online Reading Communities
- Participating in Virtual Book Clubs
- Following Authors and Publishers Syndrom D
- 9. Balancing eBooks and Physical Books Syndrom D
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Syndrom D
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Syndrom D
 - Setting Reading Goals Syndrom D
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Syndrom D
 - Fact-Checking eBook Content of Syndrom D
 - 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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