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Treacher Collins Syndrome

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Abstract

Treacher Collins syndrome, also known as mandibulofacial dysostosis, is a rare genetic disorder characterised by craniofacial abnormalities resulting from mutations affecting facial bone development. The condition primarily involves underdevelopment of the cheekbones, jaw, and chin, and is frequently associated with ear and eye anomalies. These structural defects can lead to significant functional challenges, including breathing difficulties, feeding problems, and hearing impairment. Diagnosis is usually based on clinical features and confirmed through genetic testing. Management of Treacher Collins syndrome requires a multidisciplinary approach involving specialists from paediatrics, otolaryngology, maxillofacial surgery, audiology, speech therapy, and nursing care. Treatment is largely supportive and may include surgical correction of facial deformities, the use of hearing aids, and interventions for speech and feeding. With early diagnosis and timely, coordinated care, individuals with Treacher Collins syndrome can achieve favourable functional outcomes and improved quality of life.

Keywords: Treacher-Collins syndrome, mandibulofacial dysostosis, multidisciplinary, genetic counselling, otolaryngology.

Introduction

Treacher Collins syndrome (TCS), also known as mandibulofacial dysostosis, is an extremely rare inherited craniofacial disorder that affects the development of the face and skull. The condition primarily alters the size, shape, and position of facial structures such as the ears, eyes, cheekbones, and jaws. These anatomical abnormalities may lead to functional difficulties, including problems with feeding, breathing, and hearing, especially during infancy and early childhood.

The clinical presentation of Treacher Collins syndrome varies widely, ranging from mild features that may go unnoticed to severe deformities requiring complex medical and surgical management. Many affected children require surgical interventions to correct facial anomalies, such as cleft palate repair, and ongoing treatment for associated hearing loss. Despite these challenges, advances in medical care and a multidisciplinary treatment approach have significantly improved outcomes. With timely intervention, regular healthcare follow-ups, and supportive therapies, children with Treacher Collins syndrome can achieve normal growth, development, and a good quality of life. Early diagnosis and management remain crucial in preventing complications and reducing the need for lifelong medical care.

Definition

Treacher Collins syndrome (TCS), also known as mandibulofacial dysostosis, is a rare inherited craniofacial genetic disorder characterised by abnormal development of the facial bones, particularly the cheekbones (zygomatic bones), jaw (micrognathia), and ears. It presents with distinctive facial features such as downward-slanting palpebral fissures, external and middle ear abnormalities leading to conductive hearing loss, and may be associated with cleft palate.

Incidence

- Treacher Collins syndrome affects approximately 1 in every 50,000 children worldwide.

Etiopathogenic (Genetic) Factors of Treacher Collins Syndrome

Genetic Basis

- **TCOF1 gene**
  - Most common cause of Treacher Collins syndrome
  - Responsible for approximately **81–93%** of cases
- **POLR1C and POLR1D genes**
  - Contribute to a smaller proportion of cases
- **Treacle protein**
  - Produced by the **TCOF1 gene**
  - Essential for **ribosome biogenesis** (rRNA production)
  - Plays a critical role in cell growth, survival, and facial development

Mechanism of Disease Development

- **Reduced rRNA production**
  - Gene mutations lead to decreased rRNA synthesis
  - Results in impaired protein production
- **Cellular stress**
  - Insufficient rRNA causes stress in rapidly developing cells
  - Neural crest cells are particularly affected
- **Apoptosis (programmed cell death)**
  - Cellular stress triggers death of neural crest cells
  - Leads to **hypoplasia** (underdevelopment) of facial bones and tissues

Inheritance Patterns

- **Autosomal dominant inheritance**
  - Seen in **TCOF1 and POLR1D** mutations
  - Only one mutated gene copy is required
  - **50% chance** of transmission from an affected parent
  - May also occur due to **spontaneous (de novo) mutations**
- **Autosomal recessive inheritance**
  - Associated with **POLR1C** mutations
  - Both parents are typically asymptomatic carriers
  - **25% chance** of the disorder in each pregnancy.

Clinical manifestations

1. **Ocular (Eye) Features**

❖ Downward-slanting palpebral fissures

- Lower eyelid coloboma (notching or absence of part of the lower eyelid)
- Sparse or absent eyelashes, especially on the lower eyelid
- Possible visual complications:
- Strabismus
- Cataracts
- Partial vision loss

2. Auricular (Ear) Features

- Absent, small, or malformed external ears (**microtia**)
- Narrowed or blocked external auditory canals (atresia or stenosis)
- Middle ear ossicle malformations
- Resultant **conductive hearing loss**

3. Craniofacial (Jaw and Midface) Features

- Underdeveloped cheekbones (**malar hypoplasia**)
- Small, receding lower jaw (**mandibular micrognathia**)
- Facial asymmetry
- Functional consequences:
- Airway obstruction
- Breathing difficulty
- Feeding problems, especially in infants

4. Oral and Palatal Features

- Cleft palate
- High-arched palate
- Small mouth opening (**microstomia**)
- Speech and feeding difficulties

5. Dental Features

- Missing teeth (**hypodontia**)
- Malformed or misaligned teeth
- Delayed eruption of teeth

6. Neurological and Developmental Features

- **Normal intelligence** in most individuals
- Normal cognitive development

7. Other Associated Features (Rare)

- Occasional involvement of other systems:
- Cardiac anomalies
- Renal abnormalities
- Limb defects

Diagnostic evaluation

1. Clinical Examination

- Presence of characteristic craniofacial features:
- Downward-slanting palpebral fissures
- Underdeveloped cheekbones (**zygomatic hypoplasia**)
- Small, receding jaw (**mandibular micrognathia**)
- Small, malformed, or absent external ears (**microtia**)

- Absent or narrowed external auditory canals
- Associated clinical findings:
- Cleft palate or high-arched palate
- Feeding difficulties
- Hearing impairment (usually conductive)

2. Imaging Studies

- **X-rays and CT scans**
- Demonstrate hypoplasia of facial bones (mandible, zygoma)
- Assess middle ear ossicles and airway
- Aid in surgical planning
- **Prenatal ultrasonography**
- Detects facial abnormalities during the **second trimester**
- Suggests diagnosis in severe cases

3. Genetic Testing

- Identification of pathogenic mutations in:
- **TCOF1 gene** (most common)
- **POLR1C or POLR1D genes** (less common)
- Confirms the clinical diagnosis and helps determine inheritance pattern

4. Associated Multidisciplinary Evaluations

- **Audiological assessment**
- Evaluation of conductive hearing loss
- Structural assessment of middle and inner ear
- **Ophthalmological evaluation**
- Detection of eyelid coloboma and other ocular abnormalities
- **Speech and feeding assessment**
- Evaluation of difficulties related to cleft palate or micrognathia
- **Genetic counseling**
- Explanation of inheritance patterns
- Risk assessment for future pregnancies

Management

Goal: To enhance function and quality of life, as there's no cure.

1. Airway Management

- Primary and immediate priority, especially in neonates
- Management options include:
  - Specialized positioning and intubation
  - Tracheostomy in severe airway obstruction
- Ensures safe breathing and anesthesia during procedures

2. Feeding and Swallowing Management

- Management of feeding difficulties due to cleft palate and micrognathia
- Interventions include:
  - Surgical repair of cleft palate

- Use of specialized feeding techniques
- Gastrostomy tube (G-tube) feeding when necessary

3. Management of Hearing Loss

- Early assessment and intervention for conductive hearing loss
- Treatment options:
  - Hearing aids
  - Bone conduction devices (e.g., BAHA)
  - Cochlear implants in selected cases
- Usually introduced after early childhood to support speech development

4. Craniofacial Reconstruction

- Staged surgical procedures tailored to growth and development
- Includes:
  - Zygomatic (cheekbone) reconstruction
  - Mandibular distraction osteogenesis
  - Orthognathic surgery in adolescence

5. Ear Reconstruction

- Surgical creation of anatomically typical external ears
- Uses costal cartilage grafts or synthetic implants
- Commonly performed between **6–8 years of age**

6. Dental and Orthodontic Care

- Long-term orthodontic management
- Correction of jaw alignment and dental anomalies as facial bones grow

7. Ophthalmic Care

- Management of lower eyelid coloboma and eye exposure
- Prevents corneal dryness, irritation, and damage

8. Speech and Language Therapy

- Improves articulation and oral motor skills
- Especially important in children with cleft palate and hearing loss

9. Psychosocial Support

- Psychological counseling and social support
- Helps address self-esteem, emotional stress, and social integration

Multidisciplinary Team Approach

- Effective management requires a specialised **craniofacial team**, including:
  - Plastic, maxillofacial, and neurosurgeons
  - ENT specialists
  - Audiologists
  - Dentists and orthodontists
  - Nurse
  - Speech and language therapists
  - Geneticists
  - Psychologists and social workers

Prognosis / Outlook

- With early diagnosis and consistent multidisciplinary care:
  - Individuals with Treacher Collins syndrome often lead **long, healthy lives**
  - **Intelligence is usually normal**
  - Treatment focuses on optimising **functional abilities and facial aesthetics**

Life Expectancy of Treacher Collins Syndrome (TCS)

Treacher Collins syndrome does **not usually affect life expectancy**. Most individuals with TCS have a **normal lifespan**, especially when the condition is diagnosed early and managed appropriately.

Key points:

- **Intelligence is typically normal**, and the disorder mainly affects craniofacial structures.
- **Life-threatening complications**, when they occur, are usually related to:
  - Severe airway obstruction in infancy
  - Feeding difficulties
  - Recurrent respiratory infections
- With **early airway management**, proper feeding support, and multidisciplinary medical care, these risks are greatly reduced.
- Advances in **neonatal care, surgical techniques, hearing rehabilitation, and long-term follow-up** have significantly improved outcomes.
- With timely intervention and ongoing healthcare support, individuals with Treacher Collins syndrome can expect to live **long, healthy, and productive lives**, comparable to the general population.

Preventive Factors of Treacher Collins Syndrome (TCS)

Treacher Collins syndrome is a **genetic disorder** and therefore **cannot be completely prevented**. However, certain measures can help **reduce risk, enable early detection, and prevent complications**.

1. Genetic Counselling

- Recommended for individuals or families with a history of TCS

- Helps assess:
  - Inheritance patterns (autosomal dominant or recessive)
  - Risk of transmission to future offspring
- Assists informed family planning decisions

2. Prenatal Screening and Diagnosis

- **Prenatal ultrasound** can detect severe craniofacial anomalies during the second trimester
- **Prenatal genetic testing** (when there is known family history) can identify mutations in:
  - TCOF1
  - POLR1C
  - POLR1D

3. Avoidance of Non-Genetic Risk Factors

- Although TCS is not caused by environmental factors, maintaining **good maternal health** supports normal fetal development:
  - Avoidance of alcohol, smoking, and teratogenic drugs during pregnancy
  - Proper prenatal nutrition and care

4. Early Postnatal Intervention (Secondary Prevention)

- Early identification allows prevention of complications:
  - Prompt airway management
  - Early feeding support
  - Hearing screening and intervention
- Prevents long-term complications and developmental delays

5. Regular Follow-up and Monitoring

- Ongoing assessments by a multidisciplinary team
- Early detection of:
  - Airway compromise
  - Hearing loss
  - Speech and dental issues

Conclusion

Treacher Collins syndrome is a rare genetic craniofacial disorder caused by mutations affecting facial development. Although it presents with significant structural and functional challenges, intelligence is usually normal. Early diagnosis and multidisciplinary management are crucial in preventing complications and improving quality of life. With timely intervention and continuous care, individuals with Treacher Collins syndrome can lead healthy and productive lives.

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