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Review Article

Treacher Collins syndrome: current and emerging treatment options

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ABSTRACT

Treacher Collins syndrome is a rare congenital disorder characterized by bilateral craniofacial malformations, including mandibular and zygomatic hypoplasia, cleft palate, and eye anomalies, resulting in significant variability in symptom severity. The syndrome, with an incidence of approximately 1 in 50,000 births, is primarily caused by mutations in the TCOF1 gene and less frequently by mutations in POLR1C and POLR1D genes, disrupting ribosome biogenesis crucial for craniofacial development. Clinical manifestations encompass diverse craniofacial, auditory, and skeletal abnormalities, often leading to airway complications and hearing loss. Management involves a multidisciplinary approach, combining surgical interventions such as mandibular distraction osteogenesis and orthognathic surgeries with advanced imaging techniques for precise planning. Hearing loss is managed with bone conduction aids and cochlear implants, although outcomes may vary due to associated anomalies. Genetic testing aids in diagnosis and family planning decisions, while emerging therapies like genome editing and regenerative medicine show promise for future treatments. Despite challenges in long-term management and relapse rates in surgical corrections, ongoing research aims to enhance therapeutic efficacy and patient outcomes, emphasizing the need for tailored treatment strategies addressing both functional and aesthetic aspects.

Keywords: Craniofacial malformations, Genetic mutations, Multidisciplinary approach, Regenerative medicine, Surgical intervention, Treacher Collins syndrome

INTRODUCTION

Treacher Collins syndrome (TCS) is a rare congenital craniofacial disorder characterized by distinctive bilateral craniofacial malformations, including mandibular hypoplasia, zygomatic hypoplasia, cleft palate, and palpebral fissures, often leading to difficulties in airway management and hearing loss.[1] The syndrome exhibits high phenotypic variability and incomplete penetrance, with some patients experiencing severe craniofacial deformities and others presenting milder symptoms such as hearing loss or auricle dysplasia.[2] It has an incidence rate of approximately 1 in 50,000 live births.[3] Is primarily caused by mutations in the TCOF1 gene, which is located in chromosome, encodes the nucleolar protein treacle, essential for ribosomal RNA (rRNA) synthesis, and in the POLR1C and POLR1D genes, which are crucial for RNA polymerase I and III transcription.[1,5]

These mutations disrupt ribosome biogenesis, leading to oxidative stress, DNA damage, and increased cell death in neural crest cells, which are vital for craniofacial development.[4] The mutations in the TCOF1 gene, accounts for over 90% of cases, while mutations in

POLR1C and POLR1D genes are responsible for a smaller percentage.[5]

CLINICAL FEATURES, INCLUDING CRANIOFACIAL, AUDITORY AND SKELETAL MANIFESTATIONS.

Typical craniofacial attributes consist of colobomas in the lower eyelids, palpebral fissures that slant downwards, mandibular hypoplasia, underdeveloped zygomatic bones and micrognathia. Patients often present with malformed pinnae and auricle dysplasia, which can lead to conductive hearing loss due to anomalies such as fusion of the malleus and incus, sclerotic mastoid, and hypoplastic middle ear space.[6] Bone manifestations include short anterior and posterior cranial bases, and in some cases, monopodial stapes and pectus carinatum have been reported.[7] Prenatal ultrasound can detect TCS features such as mandibular hypoplasia and bilateral auricle defects are often visible by the early second trimester.

Impact on the quality of life of patients

TCS significantly impacts patients' quality of life (QoL) across various domains, including physical health, psychological well-being, and social functioning. Individuals with TCS often experience obstructive sleep apnea (OSA), which is prevalent in 87% of patients and is associated with poorer health-related quality of life (HRQoL) due to factors like increased wake time after sleep and subjective snoring.[8] Additionally, adults with TCS report the highest levels of depression and the lowest levels of overall QoL, well-being, and physical health-related QoL compared to those with other orofacial conditions like cherubism and oligodontia/ectodermal dysplasia (ED).[9] Common functional impairments include snoring, impaired phonation, and nasal obstruction, often exacerbated by septal deviations and external nasal deformities. Surgical treatments, such as orthognathic surgery, aim to enhance occlusal function and facial aesthetics. Despite improvements, there may be some recurrence of symptoms and lingering dissatisfaction with facial features such as ears and eyelids. Despite these challenges, patients generally maintain normal intelligence, although learning disabilities are common in early life due to hearing loss and other developmental abnormalities.[10]

GENETICS OF TREACHER COLLINS SYNDROME

Patterns of inheritance

Is primarily inherited in an autosomal dominant manner although, there are rare instances of autosomal recessive inheritance, particularly involving mutations in POLR1C and POLR1D.[11,12] Molecular studies have shown that TCOF1 mutations often lead to premature protein termination, and gene rearrangements account for a significant proportion of TCS cases, indicating the importance of dosage analysis alongside sequence analysis for accurate diagnosis.[13]

Genetic mutations associated

Notably, mutations in TCOF1 are associated with a wide range of phenotypic variability, including in some cases, intellectual disability, particularly when large deletions encompass additional genes such as CAMK2A or SLC6A7. Studies have identified specific mutation hotspots within these genes, such as exon 24 in TCOF1 and exons 3, 8, and 15 in POLR1D, POLR1C, and POLR1B, respectively, with certain variants correlating with higher severity levels, particularly in Asian populations.[1,14] Additionally, novel mutations continue to be discovered, expanding the mutationspectrum, and enhancing the understanding of the genotype-phenotype correlations in TCS.

DIAGNOSIS OF TREACHER COLLINS SYNDROME

Diagnostic evaluation

The diagnosis is primarily clinical, based on characteristic facial features such as antimongoloid slanting of the palpebral fissures, coloboma of the lower eyelid, and down slanting palpebral fissures, which can be detected as early as the second trimester via prenatal ultrasound.[7,15] Genetic testing is crucial for confirming the diagnosis. Cephalometric analysis and three-dimensional craniometric analyses are valuable for assessing the severity of craniofacial deformities and planning surgical interventions, as they reveal significant deviations from normative data in mandibular projection, maxillary position, and nasal airway volume.[16,17]

Surgical planning for TCS often involves geometric morphometrics to guide orbitozygomatic and mandibular reconstruction, ensuring precise correction of skeletal deformities. Additionally, bone conduction hearing rehabilitation is recommended for patients with bilateral conductive hearing loss, showing significant improvements in hearing and speech discrimination.[3] Differential diagnosis is essential, as TCS shares clinical features with other syndromes like otodental syndrome, necessitating genetic analysis for accurate diagnosis. Overall, a multidisciplinary approach involving genetic testing, detailed clinical evaluation, and advanced imaging techniques is essential for the effective diagnosis and management.

Genetic testing

Next-generation sequencing (NGS) and Sanger sequencing are commonly used to identify these mutations, as demonstrated in studies where novel variants in TCOF1 were discovered, expanding the

mutation spectrum and aiding in prenatal diagnosis and fertility guidance for affected families. Additionally, whole-exome sequencing (WES) combined with Sanger sequencing has proven effective in identifying pathogenic variants in TCS patients, enabling targeted interventions such as bone conduction hearing rehabilitation to improve quality of life.[3,11] Genetic testing not only confirms the diagnosis but also guides clinical management and provides valuable information for family planning and early intervention strategies.

Prenatal diagnosis

Prenatal diagnosis of TCS involves a combination of genetic testing and imaging techniques to identify characteristic features and mutations associated with the disorder. Molecular genetic testing can be performed in the first trimester to predict the likelihood of the fetus being affected.[18] Ultrasound imaging plays a vital role, often complementing genetic testing, in identifying physical anomalies such as mandibular and zygomatic hypoplasia, malformed pinnae, and other craniofacial abnormalities commonly seen in TCS. In some cases, severe craniofacial anomalies, including agenesis of nasal passages and abnormal maxilla development, can be detected, which are associated with increased apoptosis in neural crest cells due to treacle haploinsufficiency.[19,20]

CURRENT MANAGEMENT OF TREACHER COLLINS SYNDROME

Multidisciplinary approach

The management of TCS involves a team of specialists such as craniofacial surgeons, pediatricians, geneticists, and anesthesiologists. Surgical interventions often include mandibular distraction, zygomatic reconstruction, and cleft palate repair, which necessitate precise preoperative planning and the use of advanced imaging techniques like three-dimensional CT scans for accurate assessment and surgical planning.[21,22]Airway management is particularly challenging in TCS patients due to anatomical abnormalities, and techniques such as fiberoptic intubation, videolaryngoscopy, and the use of supraglottic devices are often employed to ensure safe anesthesia.[23] The prevalence of difficult airway management in TCS patients is notably high, with a reported 93.3% in a study spanning 27 years, highlighting the importance of advanced airway management techniques during surgical procedures.[24]

Additionally, novel therapeutic approaches, such as the use of antioxidants to mitigate oxidative stress and the potential role of CNBP, a protein required for proper craniofacial development, are being explored to improve outcomes in TCS patients.[25] The integration of these diverse specialties and advanced techniques underscores the importance of a coordinated multidisciplinary approach in managing TCS, aiming to address both the functional and aesthetic needs of the patients while minimizing complications and improving quality of life.

Aesthetic and functional surgery

Surgical interventions are crucial for correcting facial deformities and improving airway patency. Techniques such as counterclockwise craniofacial distraction osteogenesis can address both facial support and airway issues by moving the entire facial skeleton rather than just the mandible.[26] Orthognathic surgeries, including Le Fort I and bilateral sagittal split osteotomies, are often required to correct malocclusion and improve facial aesthetics, with long-term stability reported in bimaxillary surgeries.[27] Mandibular Distraction Osteogenesis (MDO) has been used to address severe airway obstructions and facilitate tracheostomy removal, although its success varies between TCS and other conditions like Robin Sequence.[28] Additionally, a combination of chin advancement, malar osteotomies, and maxillomandibular osteotomies has been employed to correct facial deficiencies, with the flexibility to adapt to individual patient needs.[29]

Additionally, bone conduction hearing rehabilitation and zygomatic reconstruction using calvarial bone grafts are common procedures to address hearing loss and facial asymmetry.[30] Orthodontic treatment is essential for managing occlusal relationships and often involves pre and postsurgical orthodontic care to ensure optimal dental alignment and function. Facial therapy, encompassing manual orofacial techniques and alternative feeding approaches, is essential for enhancing feeding skills and mitigating risks such as failure to thrive and repeated aspirations in infants.

Management of hearing loss, including hearing aids and cochlear implants

Management of hearing loss in TCS involves a multifaceted approach, including the use of hearing aids and cochlear implants. Bone conduction hearing aids can significantly improve hearing by bypassing the outer and middle ear malformations. Cochlear implants are another viable option, especially for those with severe to profound hearing loss.31 However, the efficacy of cochlear implants can be affected by the presence of concurrent anomalies such as cochlear nerve deficiency (CND), necessitating comprehensive preoperative evaluation using MRI and other diagnostic methods to determine the suitability of cochlear implants or auditory brainstem implants (ABI).32 Moreover, the decision-making process regarding interventions like cochlear implants should take into account factors such as the patient's age, cognitive development, and potential for auditory rehabilitation, given the considerable variability in outcomes. Postoperative care and rehabilitation play a critical role in maximizing the advantages of cochlear implants and may involve auditory training and counseling to address both peripheral and central auditory

processing challenges. The development of self-administered surveys aimed at assessing cochlear implant management skills can assist in monitoring and enhancing patient outcomes.

Genetic counseling and psychosocial support

Genetic counselors must address psychological defense mechanisms like denial and grief in affected individuals and their families to facilitate informed reproductive decisions and provide holistic support.[33] Additionally, the identification of TCS through genetic testing allows for early intervention and personalized care, impacting clinical practice positively. Understanding the genetic basis of TCS aids in developing therapeutic interventions and preventive strategies, emphasizing the importance of genetic counseling in the multidisciplinary management of this syndrome.

Genome editing

Genome editing technologies, such as CRISPR-Cas9, hold potential for correcting genetic mutations at the DNA level. WES combined with Sanger sequencing has been effective in identifying novel TCOF1 mutations, which could be targeted by genome editing tools.[3] However, the complexity of TCS, including the presence of partial gene deletions and the need for precise dosage of treacle, poses challenges for genome editing. Despite these challenges, advancements in genome editing and a deeper understanding of the molecular mechanisms underlying TCS could pave the way for potential therapeutic interventions, offering hope for improved outcomes in patients with this syndrome.

Regenerative medicine

While current approaches like mandibular distraction osteogenesis and orthognathic surgery effectively enhance airway function and facial morphology, they typically entail intricate, invasive procedures that carry notable risks of relapse. Regenerative techniques, such as the use of bone marrow mesenchymal stem cells (BMMSCs) and adipose-derived mesenchymal stem cells (ADSCs), have been explored for craniofacial regeneration, but their ability to fully replicate the properties of cranial neural crest cells (CNCCs) remains limited.[34] Recent studies using zebrafish models have demonstrated that restoring the expression of key genes like POLR1C and TCOF1 can rescue craniofacial malformations by correcting neural crest cell expression and reducing cell death.35 Additionally, the use of antioxidants and overexpression of CNBP has shown potential in ameliorating craniofacial defects by reducing oxidative stress and supporting cell survival.[4,25] Despite these advances, the clinical application of regenerative medicine in TCS is still in its infancy, with more research needed to refine these techniques and ensure their safety and efficacy in human patients. The development of engineered cartilage and the use of periosteal bone flaps have also been explored as potential regenerative strategies, offering less invasive alternatives to traditional surgical methods.[36] Overall, while regenerative medicine offers a promising avenue for treating TCS, further studies are required to translate these findings into effective clinical therapies.

New therapeutic approaches

Recent therapeutic approaches have focused on both surgical and genetic interventions. Genioplasty distraction combined with hyoid advancement has shown promise in optimizing epiglottal positioning and resolving obstructive sleep apnea.[37] Additionally, virtual normalization using geometric morphometrics provides a standardized tool for planning orbitozygomatic and mandibular reconstruction, enhancing surgical precision.[22] On the genetic front, inhibition of p53 has been identified as a potential therapeutic avenue, as it prevents neuroepithelial apoptosis and rescues craniofacial abnormalities in animal models, suggesting a possible clinical application for preventing TCS birth defects.[38] Despite these advancements, long-term stability of orthognathic corrections remains a concern, with significant relapse observed in some cases, necessitating ongoing orthodontic management.[27] The combination of these innovative surgical techniques and genetic interventions holds promise for improving outcomes in TCS patients, although individualized treatment plans are essential due to the syndrome's phenotypic variability.

CONCLUSIONS

TCS presents a complex clinical landscape. Management necessitates a multidisciplinary approach encompassing surgical interventions, orthodontics, and hearing rehabilitation tailored to individual needs. Advances in surgical techniques like distraction osteogenesis and genetic insights from genome editing offer potential avenues for improving outcomes. However, challenges persist, including the risk of relapse post-surgery and variability in response to interventions like cochlear implants. Psychosocial support and genetic counselling are integral, aiding families in navigating the complexities of diagnosis and management decisions. Regenerative medicine holds promise but requires further research to refine techniques for clinical application effectively. Overall, while strides have been made in understanding and managing TCS, continued multidisciplinary collaboration and innovative approaches are essential to enhance care and quality of life for affected individuals.

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