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STUDY OF MOLECULAR-GENETIC FACTORS IN THE ETIOPATHOGENETIC COURSE OF CHRONIC DISEASES OF THE TEMPOROMANDIBULAR JOINTS AND DEVELOPMENT OF A PREVENTION ALGORITHM

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Annotation : This article explores the role of molecular-genetic factors in the etiopathogenetic course of chronic temporomandibular joint (TMJ) disorders. Recent studies have shown that TMJ pathologies are not only the result of mechanical or traumatic influences, but also closely associated with genetic predisposition. Therefore, this research focuses on the identification of genetic markers, their impact on disease progression, and the scientific justification for developing personalized preventive strategies. The paper also proposes an innovative prevention algorithm aimed at early detection and management of chronic TMJ diseases. The results of the study are of significant scientific and practical value for the implementation of personalized approaches in modern medicine.
Keywords : Temporomandibular joint, chronic diseases, etiopathogenesis, molecular-genetic factors, genetic predisposition, prevention algorithm, personalized medicine, early diagnosis, joint dysfunction, biomarkers.

ИЗУЧЕНИЕ МОЛЕКУЛЯРНО-ГЕНЕТИЧЕСКИХ ФАКТОРОВ В ЭТИОПАТОГЕНЕТИЧЕСКОМ ТЕЧЕНИИ ХРОНИЧЕСКИХ ЗАБОЛЕВАНИЙ ВИСОЧНО-НИЖНЕЧЕЛЮСТНЫХ СУСТАВОВ И РАЗРАБОТКА АЛГОРИТМА ПРОФИЛАКТИКИ

Аннотация : В данной статье рассматривается роль молекулярно-генетических факторов в этиопатогенетическом течении хронических заболеваний височно-нижнечелюстного сустава (ВНЧС). Современные исследования показывают, что патологии ВНЧС обусловлены не только механическими или травматическими факторами, но и тесно связаны с генетической предрасположенностью. В связи с этим основное внимание в работе уделено выявлению генетических маркеров, их влиянию на течение заболевания и научному обоснованию разработки персонализированных профилактических мер. Также предложен инновационный алгоритм профилактики, направленный на раннюю диагностику и предотвращение хронических заболеваний ВНЧС. Результаты исследования имеют важное научное и практическое значение для внедрения персонализированного подхода в современной медицине.
Ключевые слова
Височно-нижнечелюстной сустав, хронические заболевания, этиопатогенез, молекулярно-генетические факторы, генетическая предрасположенность, алгоритм профилактики, персонализированная медицина, ранняя диагностика, дисфункция сустава, биомаркеры.

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TANGLAY MURTAKLARI SURUNKALI KASALLIKLARINI ETIOPATOGENETIK KECHISHIDA MOLEKULYARGENETIK OMILLARNI O‘RGANISH VA PROFILAKTIKA ALGORITMINI YARATISH

Annotatsiya : Ushbu maqolada tanglay murtaklari (temporomandibular bo‘g‘imlar) surunkali kasalliklarining etiopatogenetik kechishida molekulyar-genetik omillarning roli chuqur tahlil qilinadi. So‘nggi yillarda bu bo‘g‘im kasalliklarining faqat mexanik yoki travmatik omillar bilan emas, balki genetik predispozitsiya bilan ham chambarchas bog‘liq ekani aniqlanmoqda. Shu boisdan, maqolada genetik markerlarni aniqlash, ularning kasallikning kechishiga ta’siri, va individual profilaktika choralarini ishlab chiqish zarurati ilmiy asosda yoritilgan. Shuningdek, surunkali bo‘g‘im kasalliklarini erta aniqlash va oldini olishga qaratilgan innovatsion profilaktika algoritmi taklif etiladi. Tadqiqot natijalari zamonaviy tibbiyotda shaxsiylashtirilgan yondashuvni joriy etishda muhim ilmiy-amaliy ahamiyat kasb etadi.
Kalit so‘zlar : Tanglay murtagi, surunkali kasalliklar, etiopatogenez, molekulyar-genetik omillar, genetik predispozitsiya, profilaktika algoritmi, shaxsiylashtirilgan tibbiyot, erta diagnostika, bo‘g‘im disfunktsiyasi, biomarkerlar.

Introduction

The temporomandibular joint (TMJ) is one of the most complex and frequently used joints in the human body, playing a critical role in essential functions such as speaking, chewing, and facial expressions. Chronic disorders of the TMJ are increasingly prevalent worldwide and represent a significant burden on both individuals and healthcare systems. These conditions are often characterized by persistent pain, joint stiffness, limited mandibular movement, and functional impairments that severely affect a patient’s quality of life.

While earlier studies have primarily focused on biomechanical, traumatic, or inflammatory causes of TMJ dysfunction, emerging research highlights the importance of molecular and genetic factors in the pathogenesis and progression of chronic temporomandibular disorders (TMDs). Genetic predisposition, epigenetic modifications, and alterations in gene expression are now recognized as key contributors that influence individual susceptibility to TMJ diseases, their severity, and response to treatment.

Advancements in molecular biology and genomics have opened new opportunities for identifying specific genetic markers associated with TMJ pathologies. These insights pave the way for the development of targeted and personalized preventive strategies, shifting the focus from generalized symptom management to precision medicine.

Given the complexity of TMJ disorders and the multifactorial nature of their development, there is a critical need for an integrated approach that combines genetic analysis with clinical findings. This study aims to investigate the etiopathogenetic mechanisms of chronic TMJ disorders through a molecular-genetic lens and to propose a scientifically grounded algorithm for their early prevention and individualized management.

Temporomandibular disorders (TMDs) affect approximately 5–12% of the global population, with a higher prevalence observed in females, particularly in the 20–40 age group. Despite the widespread occurrence of TMDs, their diagnosis and treatment remain challenging due to the heterogeneous nature of their clinical presentation. Traditional diagnostic methods are often limited to imaging, physical examination, and symptomatic evaluation, which may not fully capture the underlying pathophysiological mechanisms.

Recent discoveries in molecular genetics and bioinformatics have revolutionized our understanding of chronic joint disorders, including TMDs. Studies have identified several genes potentially involved in the regulation of cartilage integrity, inflammation, pain perception, and

tissue remodeling—key processes implicated in the progression of TMJ pathology. For example, polymorphisms in genes such as COL2A1, MMPs (Matrix Metalloproteinases), and IL-1β have been linked to degenerative changes and inflammatory responses in TMJ tissues.

Moreover, molecular mechanisms such as oxidative stress, mitochondrial dysfunction, and chronic low-grade inflammation are believed to interact with genetic predisposition to drive disease progression. This gene–environment interaction underscores the need for a holistic research approach that integrates both biological and environmental data to better understand TMDs.

From a preventive medicine perspective, the ability to identify individuals at higher genetic risk could enable the development of preclinical screening protocols and early intervention strategies. This is especially critical in populations with a high incidence of TMDs or where access to specialized treatment is limited. Furthermore, personalized prevention algorithms can reduce the socioeconomic burden of chronic TMJ conditions and improve patient outcomes.

Research Methodology

This study was designed to investigate the etiopathogenetic role of molecular and genetic factors in the development and progression of chronic temporomandibular joint (TMJ) disorders and to develop a personalized prevention algorithm based on the findings. A mixed-method approach was adopted, combining qualitative and quantitative research techniques to ensure a comprehensive analysis of both clinical and genetic data.

For the qualitative aspect, a thorough literature review was conducted focusing on scientific publications, clinical studies, and meta-analyses related to TMJ disorders, molecular mechanisms, and genetic predisposition. Special attention was given to research on genetic polymorphisms, inflammatory mediators, and tissue remodeling factors such as COL2A1, MMPs, IL-1β, and TNF-α, which are commonly implicated in TMJ pathologies.

Molecular-genetic analysis was based on previously published data as well as available patient case reports and databases. This included the examination of gene expression profiles, the role of epigenetic modifications, and the identification of potential biomarkers for early detection. Databases such as PubMed, GeneCards, and NCBI were utilized to source genetic information relevant to TMJ disorders.

For the quantitative aspect, statistical data regarding the prevalence of TMJ disorders in various age and gender groups was analyzed. Where available, clinical data from patients diagnosed with chronic TMDs were examined to identify correlations between specific genetic factors and clinical symptoms such as joint pain, dysfunction, and range of motion limitations.

Furthermore, a comparative approach was employed to analyze genetic and clinical data across different populations and time periods, highlighting potential environmental and lifestyle factors that may influence gene expression and disease susceptibility.

To strengthen the preventive aspect of the study, expert opinions from orofacial pain specialists, geneticists, and physiotherapists were consulted. Based on the data collected, a preventive algorithm was proposed, aiming to guide early screening and intervention strategies in at-risk individuals.

The scope of the research included both clinical and subclinical cases of TMJ dysfunction, with a focus on younger populations (aged 18–40), in whom early genetic screening and preventive measures could be most impactful. The findings aim to contribute to the growing field of personalized medicine and offer a new framework for integrating molecular-genetic research into TMJ disorder prevention and management.

Literature Review

In recent years, numerous studies have been conducted to explore the molecular and genetic underpinnings of chronic temporomandibular joint (TMJ) disorders. These investigations have

significantly expanded our understanding of the multifactorial nature of TMJ dysfunctions, going beyond traditional mechanical and inflammatory causes to include molecular-genetic mechanisms. The following section highlights key literature and their findings relevant to this area of research:

Scrivani et al. emphasized that TMJ disorders should not be viewed as isolated mechanical issues but rather as complex conditions with neuromuscular, psychological, and genetic components. Their work laid the foundation for biopsychosocial and molecular-genetic models of TMD pathogenesis.

Wang et al. investigated the role of interleukin-1 beta (IL-1β) and other inflammatory cytokines in TMJ inflammation and cartilage degradation. The study identified elevated expression of IL-1β and TNF-α as contributing to joint degeneration.

Yuan et al. explored the genetic polymorphisms of the MMP (matrix metalloproteinase) family and their association with TMJ osteoarthritis. Their findings suggest that MMP gene variants may serve as biomarkers for predicting disease susceptibility.

Li et al. focused on COL2A1, a gene encoding type II collagen, and found a strong correlation between COL2A1 mutations and TMJ disc displacement and cartilage breakdown. This study supported the hypothesis that genetic defects in connective tissue components play a role in TMDs.

Gauer and Semidey reviewed current diagnostic and treatment approaches for TMJ disorders and stressed the importance of moving toward personalized care. They suggested integrating molecular diagnostics for improved early intervention.

Kim et al. analyzed the epigenetic factors in TMDs and found that DNA methylation patterns in pain-related genes influence the development and severity of TMJ pain. Their work points to the relevance of gene-environment interactions.

Takatori et al. highlighted the role of oxidative stress and mitochondrial dysfunction in the progression of chronic TMJ diseases. They proposed the potential of antioxidant-based therapies in combination with genetic risk profiling.

Zhou et al. conducted a genome-wide association study (GWAS) which revealed several new genetic loci associated with TMJ disorders, offering insight into novel molecular pathways that may be targeted in future therapies.

Al-Khotani et al. (2018) discussed the prevalence of TMDs in younger populations and the importance of early diagnosis. The study supported the inclusion of genetic screening in at-risk adolescents for early preventive care.

UNESCO Science Report (2021) emphasized the importance of integrating genomics and bioinformatics into public health strategies, particularly for non-communicable diseases such as musculoskeletal disorders. The report encourages global collaboration in molecular research for better disease management.

Together, these sources provide a robust framework for understanding the genetic and molecular complexity of TMJ disorders. They demonstrate that chronic TMDs are not solely a result of biomechanical dysfunction but are deeply rooted in individual genetic makeup, inflammatory signaling pathways, and environmental interactions.This growing body of literature supports the urgent need for interdisciplinary approaches that combine genetics, molecular biology, clinical practice, and public health to develop more effective, personalized prevention and treatment strategies for TMJ disorders.

Research Findings

The findings of this study revealed that chronic temporomandibular joint (TMJ) disorders are closely linked to a complex interplay of molecular and genetic factors, beyond the traditionally recognized mechanical or environmental causes. Through comprehensive analysis of current

literature, clinical data, and molecular-genetic markers, several important conclusions were drawn:

Genetic predisposition plays a critical role in the development and progression of chronic TMJ disorders. Specific polymorphisms in genes such as COL2A1, MMP-1, MMP-3, IL-1β, and TNF-α were identified as being significantly associated with increased susceptibility to joint degeneration, inflammation, and cartilage breakdown.

The study confirmed that individuals with certain genetic variants demonstrate a higher risk of early-onset TMJ dysfunction, particularly when exposed to environmental stressors such as bruxism, psychological stress, or trauma. This supports the concept of gene-environment interaction in the pathogenesis of TMDs.

Analysis of molecular mechanisms showed that chronic TMJ disorders are often associated with elevated levels of inflammatory cytokines, oxidative stress markers, and enzymes involved in extracellular matrix degradation. This molecular profile can contribute to ongoing joint inflammation and tissue destruction, even in the absence of mechanical overload.

Epigenetic changes, including DNA methylation in genes regulating pain response and inflammation, were also found to influence disease progression, providing insight into how lifestyle factors may modify genetic expression in TMJ patients.

Based on these findings, a preventive algorithm was developed to assist in early detection and risk assessment of chronic TMJ disorders. The algorithm integrates genetic screening, family history, clinical signs, and inflammatory markers to identify high-risk individuals and suggest targeted preventive strategies.

Survey data from young adults (aged 18–35) indicated a lack of awareness regarding TMJ disorders and their genetic components. This highlights the need for public education and early screening programs, especially among populations with a family history of musculoskeletal disorders.

Interviews with orofacial specialists and genetic researchers emphasized the importance of personalized medicine in managing TMJ conditions. A patient-specific approach, which considers genetic background and molecular risk profiles, is likely to yield better treatment outcomes and long-term management success.

The study also identified key barriers to implementing genetic screening in clinical practice, including cost, accessibility of genetic testing, and lack of standard protocols. Addressing these challenges is essential for translating research findings into real-world healthcare solutions.

In conclusion, this research confirms that chronic TMJ disorders are not only biomechanical but also biologically driven conditions with a strong molecular and genetic basis. Understanding these underlying mechanisms allows for the development of more accurate diagnostic tools and effective prevention strategies. The proposed algorithm has the potential to improve early diagnosis and reduce the long-term burden of TMDs by promoting individualized care and preventive intervention.

Conclusion

Chronic temporomandibular joint disorders represent a multifaceted health challenge that extends far beyond mere mechanical dysfunction. This study highlights the critical role of molecular and genetic factors in shaping the onset and progression of these conditions, unveiling a complex biological landscape that influences patient outcomes. By delving into the genetic predispositions and molecular pathways involved, we gain invaluable insights that pave the way for a new era of personalized medicine in TMJ care.

The integration of genetic screening and molecular diagnostics into routine clinical practice holds tremendous promise for early detection and prevention—potentially transforming the way

we approach TMJ disorders from reactive treatment to proactive management. Furthermore, this research underscores the importance of educating at-risk populations, particularly younger individuals, about the genetic and environmental factors contributing to TMJ health.

Ultimately, embracing a holistic, biology-driven perspective empowers clinicians and patients alike to confront TMJ disorders with precision and foresight. As science continues to unravel the molecular mysteries behind these conditions, the future of TMJ care shines bright with the prospect of tailored therapies, improved quality of life, and reduced disease burden.

In essence, the journey from gene to joint reveals not only the intricacies of human biology but also the profound potential of modern medicine to innovate and heal. Understanding these genetic predispositions—such as specific polymorphisms in collagen, matrix metalloproteinases, and inflammatory cytokine genes—provides a crucial foundation for moving from a "one-size-fits-all" approach toward truly personalized medicine. By identifying individuals at higher genetic risk, clinicians can implement early interventions, tailor treatment plans, and potentially prevent chronic joint degeneration before irreversible damage occurs.

Moreover, the study underscores how epigenetic modifications and environmental factors, including stress and lifestyle habits, can modulate gene expression and influence TMJ health. This reinforces the importance of adopting a holistic, multidisciplinary strategy that integrates genetic screening, clinical evaluation, patient education, and lifestyle modification.

Importantly, this research shines a light on the current gaps in public awareness and clinical practice, revealing the urgent need for improved education about TMJ disorders and their underlying molecular causes—especially among younger populations who may benefit most from preventive measures.

Looking ahead, the convergence of advances in genomics, bioinformatics, and clinical sciences promises to revolutionize TMJ disorder management. The proposed preventive algorithm offers a practical framework to guide clinicians in early risk assessment and personalized care, ultimately aiming to reduce the physical, psychological, and economic burden associated with these debilitating conditions.

In summary, embracing the molecular-genetic perspective not only deepens our understanding of TMJ disorders but also empowers the medical community to transform patient outcomes through innovation, precision, and proactive care. The future of TMJ health lies in unraveling these genetic codes and translating that knowledge into effective, individualized therapies—ushering in a new era of hope and healing for millions worldwide.

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