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Genetics of Inflammatory Bowel Disease: Current Understanding and Future Directions

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Aim: to present literature data on the genetic underpinnings of inflammatory bowel disease (IBD), focusing on key genetic variants such as NOD2, ATG16L1, and IL23R; The objective of this was to understand how these genetic factors contribute to immune dysregulation, epithelial barrier dysfunction, and mucosal homeostasis in IBD.
Key points. IBD, encompassing Crohn's disease and ulcerative colitis, is intricately shaped by a nexus of genetic and environmental factors, and its global prevalence is increasing. Genetic research has identified pivotal variants — NOD2, ATG16L1, and IL23R — linked to IBD, influencing key pathways, such as autophagy, interleukin signaling, and bacterial management. These genetic variants, integral to IBD's etiology of IBD, have functional significance because they perturb immune regulation, compromise epithelial barrier integrity, and disrupt mucosal homeostasis, collectively contributing to intestinal inflammation through diverse mechanisms. Early detection of IBD is paramount for arresting disease progression and underscoring the importance of prognostic testing and genetic screening, especially in cases with familial predispositions or very early onset IBD. Additionally, the use of certain IBD medications, such as corticosteroids, azathioprine, and infliximab, may have implications for male fertility, necessitating a nuanced understanding of these potential effects for informed decision-making in IBD management. This comprehensive understanding of the genetic landscape, functional implications, and diagnostic strategies is vital for advancing personalized treatments and improving outcomes in individuals with IBD.
Conclusion. IBD is a complex gastrointestinal disorder influenced by a combination of genetic and environmental factors. Genetic variants, including NOD2, ATG16L1, and IL23R, contribute to dysregulation of immune responses, epithelial barrier function, and mucosal homeostasis. While progress has been made in understanding the genetic landscape of IBD, ongoing research is needed to elucidate the functional consequences of these variants, identify causal genes, and explore gene-environment interactions. This deeper understanding holds the potential for more precise diagnostics and personalized treatments, ultimately improving outcomes in individuals living with IBD.
Keywords: genetics, knowledge gap, genetic predisposition
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Генетика воспалительных заболеваний кишечника: современные представления и направления будущих исследований

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Цель: изучение генетических основ воспалительных заболеваний кишечника (ВЗК), уделяя особое внимание ключевым генетическим вариантам, таким как NOD2, ATG16L1 и IL23R. Задачей обзора было сформулировать, как данные генетические факторы способствуют нарушению иммунной регуляции, дисфункции эпителиального барьера и гомеостазу слизистой оболочки при ВЗК.
Основные положения. ВЗК, включающие болезнь Крона и язвенный колит, формируются под сложным влиянием генетических и экологических факторов, и распространенность данной группы патологий растет во всем мире. Генетические исследования выявили основные варианты — NOD2, ATG16L1 и IL23R, — связанные с ВЗК, влияющие на ключевые пути, такие как аутофагия, сигнализация интерлейкинов и борьба с бактериями. Эти генетические варианты, являющиеся неотъемлемой частью этиологии ВЗК, имеют функциональное значение, поскольку они нарушают иммунную регуляцию, ставят под угрозу целостность эпителиального барьера и нарушают гомеостаз слизистой оболочки, в совокупности способствуя воспалению кишечника посредством различных механизмов. Раннее выявление ВЗК имеет первостепенное значение для остановки

прогрессирования заболевания и подчеркивает важность прогностического тестирования и генетического скрининга, особенно в случаях семейной предрасположенности или очень раннего начала ВЗК. Кроме того, применение некоторых препаратов для лечения ВЗК, таких как кортикостероиды, азатиоприн и инфликсимаб, может иметь последствия для мужской фертильности, что требует детального понимания этих потенциальных эффектов для принятия обоснованных решений при лечении ВЗК. Такое всестороннее понимание генетического ландшафта, функциональных последствий и диагностических стратегий имеет решающее значение для разработки персонализированных методов лечения и улучшения результатов лечения людей с ВЗК. Несмотря на достигнутый прогресс в понимании генетического ландшафта ВЗК, необходимы дальнейшие исследования для выяснения функциональных последствий этих вариантов, выявления причинных генов и изучения взаимодействия генов и окружающей среды.

Заключение. ВЗК — это группа сложных желудочно-кишечных расстройств, на которые оказывает влияние сочетание генетических и экологических факторов. Генетические варианты, включая NOD2, ATG16L1 и IL23R, способствуют нарушению регуляции иммунных реакций, эпителиальной барьерной функции и гомеостаза слизистой оболочки. Это более глубокое понимание открывает возможности для более точной диагностики и персонализированного лечения, что в итоге улучшает результаты лечения пациентов, страдающих ВЗК.

Ключевые слова: генетика, пробел в знаниях, генетическая предрасположенность

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Introduction

A cluster of chronic inflammatory disorders predominantly affecting the gastrointestinal tract comprises inflammatory bowel disease (IBD) [1]. Crohn's disease (CD) and ulcerative colitis (UC) are the two primary subcategories of this comprehensive classification. Although they share certain clinical characteristics, these conditions display unique attributes that are influenced by a complex interplay of genetic and environmental factors [2]. As a subtype of IBD, Crohn's disease exerts a broad-ranging effect on the gastrointestinal tract, extending from the mouth to the anus, and frequently involves deep penetration into the intestinal wall. It manifests as inflammation, ulcers, and the potential for complications such as strictures and fistulas [3]. Conversely, ulcerative colitis primarily targets the colon and rectum, marked by inflammation and ulcers in the colon mucosal lining, resulting in symptoms such as diarrhea, rectal bleeding, and abdominal pain [4].

According to recent studies, the prevalence of IBD is rapidly increasing worldwide, particularly in Western countries and newly industrialized countries in Africa, Asia, South America, and other regions. For example, In Italy, the estimated prevalence of IBD increased from 80.9 per 100,000 people in 1990 to 93.8 per 100,000 people in 2017 [5]. Globally, IBD accounted for 3.32 million estimated cases in 1990 and 4.90 million cases in 2019, corresponding to an increase of 47.45 % between 1990 and 2019 [6]. The age-standardized prevalence rate of IBD in the most populous country, China, is 136.2 per 100,000 [7]. According to the European Federation of Crohn's and Ulcerative Colitis Associations (EFCCA), 10 million people worldwide live with IBD, although this number may be higher [8]. By 2030, it is predicted that over 7 million individuals in Europe and the US will have IBD [9].

Numerous studies have explored the incidence of IBD in India. A study conducted in Mumbai discovered that the incidence density of IBD was 3/1000 in the initial 10 years, 3.3/1000 between 10–20 years, and 7/1000 beyond 20 years, ultimately leading to an overall prevalence of 2.8 % [10]. In 2017, a different study documented a prevalence rate of 42.8 UC patients per 100,000 individuals [11]. A more recent 2023 study indicated that the prevalence of IBD in India was 5.4 %, with 2.2 % of cases being UC and 3.2 % being CD [12]. Additionally, a review article published in 2022 highlighted that IBD tends to be more prevalent among males compared to females in India [13].

Genetics strongly influences IBD, with an estimated heritability of 60–70 % [14]. Genetic predisposition is a significant factor, and numerous genes have been identified as being associated with IBD. These genes participate in various immune-related pathways, including the regulation of inflammation and maintenance of gut barrier function [15]. Although over 200 genes have been linked to IBD, their precise roles in the disease mechanism remain incompletely understood [16]. A critical genetic element in IBD is the *NOD2* gene, which plays a crucial role in immune response regulation and is linked to heightened susceptibility to CD and UC [17]. Another important genetic element is the *ATG16L1* gene, which encodes a protein essential for autophagy that eliminates damaged cells and is linked to a higher risk of developing Crohn's disease [18]. Despite advances

in Genome-wide Association Studies (GWAS), IBD remains a complex genetic disorder characterized by the involvement of numerous genes and intricate gene-environment interactions [19]. It is essential to recognize that while genetics play a substantial role in determining susceptibility to IBD, environmental factors such as dietary choices, smoking habits, microbial exposure, and psychological stress also play pivotal roles in its initiation and progression [20]. This review will delve deeper into the genetic underpinnings of IBD and explore the latest findings and their implications for our understanding of disease mechanisms, diagnosis, and potential therapeutic interventions.

Genetic variants associated with IBD

Research into the genetic variations linked to IBD has enhanced our understanding of the underlying mechanisms of the disease. These genetic variations have been detected in individuals with IBD and are associated with autophagy, signaling pathways involving interleukin-23 and interleukin-10, impaired bacterial management, innate and adaptive immune responses, microbial recognition, and bacterial wall sensing [21]. The genetic variations associated with IBD, encompassing frequently and infrequently occurring variants, are well documented. Some widely recognized genetic variations in IBD involve genes responsible for encoding *IL23R, ATG16L1*, and *NOD2* [22], as shown in the Figure.

The prevalence of these variants varies among populations but is typically limited. For instance, *NOD2* variants are found in approximately 5 % of individuals of European descent with IBD and are associated with an elevated risk of developing CD [23]. In individuals with IBD, *IL23R* variants are present and are linked to a protective effect against CD and UC [24]. *ATG16L1* variants, found in approximately 5–10 % of individuals with IBD, are associated with an elevated likelihood of developing CD [25].

In addition to these common variants, several rare genetic variants have also been implicated in IBD. Research into rare variants of IBD faces limitations owing to the expensive and intricate nature of sequencing. Nevertheless, a few studies have indicated a connection between uncommon variants in genes, such as *NCF4, XIAP, CYBB*, and IBD, implying that these variants might exert a more substantial influence than common variants [26]. Uncommon IL10 receptor gene (*IL10R*) variants that disrupt its signaling pathway give rise to a condition known as very early onset IBD, which typically emerges in the initial months of life and frequently presents as severe [27].

IBD exhibits a polygenic nature, signifying that the condition is affected by numerous genetic

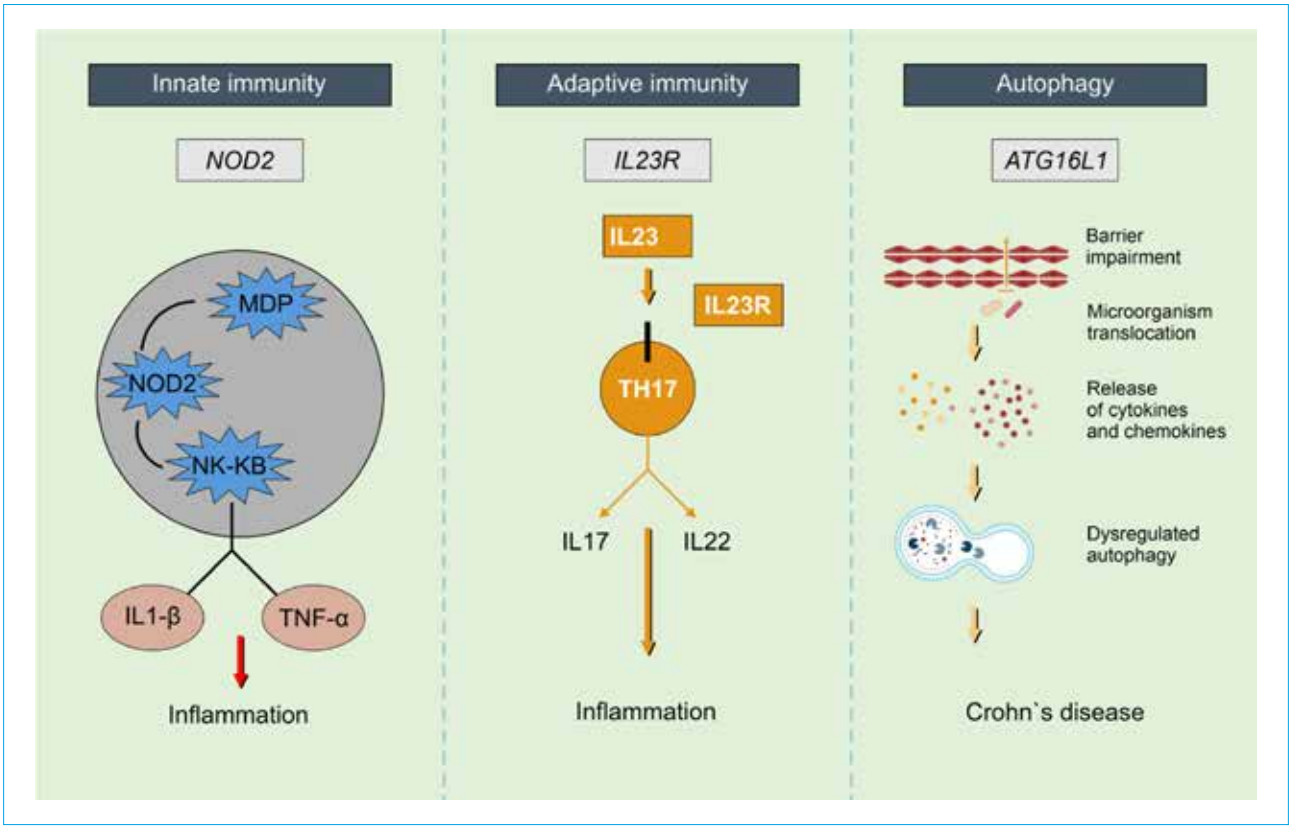

Figure. Genetic variations in IBD

Рисунок. Генетические вариации при ВЗК

variations spread across various genes, with each of these variants having a limited individual impact on the risk of developing the disease. IBD does not result from a single gene mutation or a small group of prominent genetic factors. Instead, they emerge from the cumulative impact of numerous genetic variations dispersed throughout the genome. These variations can influence diverse aspects of the inflammatory response, immune system, and intestinal barrier function [28]. The genetic framework of IBD is complex. Although specific genes, such as *NOD2, ATG16L1*, and *IL23R*, have been recognized as risk factors, the collective influence of all identified genetic variants explains only a fraction of the overall disease susceptibility [29] (Table).

NOD2 (Nucleotide-binding oligomerization domain 2)

NOD2 plays a central role in the pathophysiology of IBD, especially in CD. It is a vital component of the innate immune system and serves as the primary safeguard against pathogens. *NOD2* is primarily expressed in specialized immune cells, including macrophages, monocytes, and Paneth cells, in the small intestine [30]. *NOD2* operates as a pattern recognition receptor, capable of identifying specific elements within bacterial cell walls, particularly a molecule called muramyl dipeptide (MDP). Upon detection of MDP, *NOD2* triggers a signaling cascade, triggering a sequence of events that activate the nuclear factor-kappa B (NF-κB) and mitogen-activated protein kinase (MAPK) pathways [31]. These pathways play a central role in governing the immune response and triggering the generation of pro-inflammatory cytokines, such as interleukin-1β (IL-1β) and tumor necrosis factor-alpha (TNF-α), both of which are essential mediators of inflammation and immune reactions [32]. In CD, mutations in *NOD2* are linked to distinct clinical features, which include inflammation primarily targeting the ileum (the terminal portion of the small intestine) and the formation of strictures or narrowing of the intestine. In addition, individuals with *NOD2* mutations are more likely to require surgical intervention [33]. Furthermore, *NOD2* significantly contributes to the maintenance of intestinal barrier integrity. *NOD2* malfunction can lead to increased intestinal permeability, allowing the passage of bacteria and antigens into the mucosa, subsequently triggering inflammation [34].

ATG16L1 (Autophagy-related 16-like 1)

ATG16L1 is paramount in the progression of IBD, particularly CD. This gene is vital in autophagy and is indispensable for preserving intestinal equilibrium and controlling immune responses [35]. Autophagy is a cellular mechanism responsible for the breakdown and recycling of cellular components including impaired organelles and proteins. It maintains cellular equilibrium and eliminates intracellular pathogens including bacteria and viruses. The *ATG16L1* gene encodes a protein known as ATG16L1 (Autophagy-related 16-like 1), which is a component of a protein complex crucial for forming autophagosomes and double-membrane structures responsible for engulfing and isolating cellular components for eventual degradation. Within the intestinal context, *ATG16L1* is significantly expressed in gut epithelial cells, comprising the inner lining of the gastrointestinal tract. These cells function as protective barriers, separating luminal contents, including microorganisms, from

Table. Genetic variants associated with IBD
Таблица. Генетические варианты, связанные с ВЗК

Gene / *Ген*	Function / *Функция*	Association with IBD / *Ассоциация с ВЗК*
IL23R	Encodes the interleukin-23 receptor *Кодирует рецептор интерлейкина-23*	Protective effect against Crohn's disease and ulcerative colitis *Защитный эффект против болезни Крона и язвенного колита*
ATG16L1	Involved in autophagy *Участвует в аутофагии*	Increased risk of Crohn's disease *Повышенный риск болезни Крона*
NOD2	Encodes a protein involved in bacterial recognition *Кодирует белок, участвующий в бактериальном распознавании*	Increased risk of Crohn's disease *Повышенный риск болезни Крона*
NCF4	Encodes a protein involved in NADPH oxidase activity *Кодирует белок, участвующий в активности НАДФН-оксидазы*	Rare variants associated with IBD *Редкие варианты, связанные с ВЗК*
XIAP	Encodes a protein involved in apoptosis *Кодирует белок, участвующий в апоптозе*	Rare variants associated with IBD *Редкие варианты, связанные с ВЗК*
CYBB	Encodes the cytochrome b-245 beta chain *Кодирует бета-цепь цитохрома b-245*	Rare variants associated with IBD *Редкие варианты, связанные с ВЗК*
IL10RA	Encodes the interleukin-10 receptor alpha chain *Кодирует альфа-цепь рецептора интерлейкина-10*	Rare variants associated with very early-onset IBD *Редкие варианты, связанные с очень ранним началом ВЗК*

the underlying tissues [36]. *ATG16L1* plays a critical role in maintaining intestinal wall integrity. It regulates the renewal of epithelial cells and reduces excessive inflammation by eliminating intracellular pathogens. When *ATG16L1* malfunctions, it can lead to barrier impairment, allowing microorganisms and antigens to penetrate intestinal mucosa. *ATG16L1*-mediated autophagy has immunomodulatory effects. It regulates the release of cytokines and chemokines, thereby influencing the immune response to pathogens and antigens in the gut. Dysregulation of autophagy can result in irregular immune activation [37]. The identified risk alleles of *ATG16L1* are associated with changes in the functionality of the ATG16L1 protein. These changes can impair autophagy, especially in the presence of bacterial pathogens. This malfunction contributes to the initiation of CD by allowing the persistence of intracellular bacteria and promoting inflammation. [38]. Impaired *ATG16L1* function can also disrupt the gut microbiome composition. Dysbiosis, which is characterized by an imbalance in the microbial community, is linked to CD and can exacerbate inflammation.

IL23R (Interleukin 23 receptor)

IL23R serves as a receptor for the cytokine interleukin-23 (IL-23), which is a central regulator of the immune system. IL-23 plays a pivotal role in stimulating and functionally activating T helper 17 (Th17) cells, a specific subgroup of $CD4^+$ T cells recognized for their capacity to generate pro-inflammatory cytokines such as interleukin-17 (IL-17) and interleukin-22 (IL-22) [39]. Following activation by IL-23, Th17 cells release pro-inflammatory cytokines such as IL-17 and IL-22. These cytokines attract immune cells to areas of inflammation, induce the production of inflammatory substances, and initiate tissue damage [40]. With the assistance of cytokines derived from Th17 cells, IL-23 undermines the integrity of the intestinal epithelial barrier. This breach permits the entry of luminal contents such as bacteria and microbial products into the underlying mucosa, thus worsening inflammation. Through Th17-derived cytokines, IL-23 can weaken the structural integrity of the intestinal epithelial barrier, promoting the infiltration of luminal contents and intensifying the inflammatory reaction [41]. The signaling pathway of IL-23 can influence the composition of the gut microbiome. Disruption of IL-23 signaling can lead to changes in the microbial population, promoting the proliferation of specific bacteria that can potentially worsen inflammation in individuals with IBD [42]. In individuals with IBD, the immune response involving IL-23 and Th17 cells is chronic and dysregulated. This persistent inflammation damages the intestinal mucosa, leading to symptoms, such as abdominal pain, diarrhea, and ulcerations [43]. Furthermore, genetic variations, including those within the *IL23R* gene, can affect an individual's responsiveness to IL-23 signaling. Protective variants of *IL23R* are associated with a reduced risk of IBD, potentially by dampening Th17 responses and pro-inflammatory cytokine production [44].

Functional implications of genetic variants

Genetic variants that have been identified can play a functional role in intestinal inflammation through diverse mechanisms. Notably, a significant portion of the causative variants linked to IBD have been observed to affect gene expression rather than actual gene function [45]. An in-depth analysis of how host genetic diversity affects intestinal gene expression in IBD revealed 1854 inflammation-associated cis-expression quantitative trait loci (eQTLs) involving 190 unique eGenes. This indicates that genetic variations can affect the expression of genes associated with intestinal inflammation, potentially playing a role in the initiation and progression of the disease [46]. This suggests that genetic variations can influence the expression of genes associated with intestinal inflammation, potentially contributing to the initiation and progression of the disease. Furthermore, gene-variant enrichment scores have been employed to assess the burden of variants in IBD and statistically distinguish between CD and UC phenotypes [47]. These scores provide a genetic framework for understanding the distinctions between the primary subtypes of IBD and can help identify the functional roles of specific genetic variations in intestinal inflammation. IBD is a multifaceted condition influenced by numerous factors, including genetic and nongenetic risk elements. The functional impact of recognized genetic variations on intestinal inflammation may be affected by their interaction with non-genetic factors such as environmental triggers or an individual's microbiome [48]. Genetic variants linked to IBD influence immune dysregulation, epithelial barrier function, and mucosal homeostasis.

Immune dysregulation

IBD tolerance breakdown occurs when the immune system loses its ability to distinguish between beneficial commensal gut microbes and potentially harmful pathogens. Specific genetic variants can disrupt this tolerance breakdown, as observed in individuals with CD who carry mutations in the *NOD2* gene [49]. When the immune system fails to tolerate commensal microbes effectively, it may mistakenly trigger immune responses against these microbes, resulting in persistent inflammation of the intestinal mucosa [50]. This constant inflammation results in tissue damage, compromises the integrity of the intestinal barrier, and causes IBD symptoms. Additionally, loss of tolerance often results in an imbalance in immune responses, marked by heightened pro-inflammatory reactions and

weakened regulatory mechanisms, further fuelling the cycle of chronic inflammation in IBD.

Epithelial barrier function

Microbial interactions represent a critical dimension of how genetic variants can affect epithelial barrier function. When genetic variations result in alterations to the makeup of the gut microbiome, it can give rise to a condition called dysbiosis. Dysbiosis involves uneven distribution within the gut microbial community [51]. Dysbiosis, which is influenced by genetic factors, can initiate gut inflammation. Inflammation can negatively impact the integrity and performance of the mucosal barrier. Inflammatory mechanisms can undermine the capacity of the barrier to efficiently maintain separation between luminal contents, including microbes, and the underlying mucosal tissue [52]. Therefore, genetic variants that influence the gut microbiome can indirectly contribute to the disruption of the epithelial barrier function, creating a complex interplay between genetic factors, microbial interactions, and mucosal health in conditions such as IBD. The genetic and transcriptomic bases of intestinal epithelial barrier dysfunction in IBD have been investigated in some studies. This study identified specific single nucleotide polymorphisms associated with intestinal barrier dysfunction, with PTGER4 being implicated in CD and HNF4A in UC [53].

Mucosal homeostasis

Certain genetic variations associated with IBD can affect critical genes involved in the production and regulation of inflammatory mediators. These variants may alter the function or expression of the enzymes and receptors responsible for the synthesis or response to these mediators [15]. When individuals with these genetic variants encounter triggers such as environmental factors or microbial stimuli, their immune cells may respond by producing inflammatory mediators in an uncontrolled or excessive manner. This dysregulated production directly results from genetic alterations [54]. Overproduction of prostaglandins and leukotrienes can disrupt the delicate balance of mucosal homeostasis. These inflammatory mediators can initiate and perpetuate inflammation within the gut, leading to tissue damage and functional impairment of the intestinal barrier [55]. Continuous and uncontrolled inflammation that arises due to the disrupted production of inflammatory mediators can contribute to the initiation and progression of IBD. This enduring inflammation is characteristic of IBD and is linked to symptoms such as abdominal pain, diarrhea, and mucosal ulcerations [3].

Early detection

Early detection of IBD is crucial for preventing disease progression in specific patients [56]. Since there is no definitive test to conclusively confirm the existence of IBD, the diagnosis relies on a combination of various tests [57]. Currently, researchers are investigating the biological substances present in an individual's blood, tissues, or other bodily fluids to determine whether they can be employed to forecast the advancement of IBD in conjunction with individual factors such as diagnosis, age, and disease location. These investigations are referred to as prognostic testing [58]. The initial diagnostic steps often involve conducting laboratory tests on the blood and stool samples. Although a blood test alone cannot provide definitive confirmation of IBD, it eliminates other conditions that may manifest similar symptoms. Typically, a small blood sample is collected to assess indications of anemia or signs of infection caused by bacteria or viruses. Additionally, stool studies may be conducted to detect signs of inflammation or infection [59]. Imaging examinations of the gastrointestinal tract identify inflammation, ulcers, and other indications of IBD. Such assessments can include X-rays, CT scans, MRI scans, and capsule endoscopy [60]. Some specific genetic mutations for early diagnosis in patients with a family history of IBD by the significance of genetic testing, particularly in cases of very early onset IBD, to identify monogenic etiologies and underlying genetic susceptibilities [61]. Next-generation sequencing has been highlighted as a valuable tool for the genetic diagnosis of IBD, particularly in cases with a strong suspicion of a particular underlying genetic etiology [62]. The diagnostic approach for investigating monogenic causes of IBD-like intestinal inflammation is largely based on restricted functional screening, and functional testing is considered valuable, especially when there is a strong suspicion of a specific underlying genetic etiology [63].

Knowledge gaps and future directions

Despite substantial advancements in the understanding of the genetic and environmental elements involved in IBD, several areas of uncertainty still require additional research efforts to fill these knowledge gaps. Although more than 240 genetic risk loci associated with IBD have been identified, our understanding of their role in disease development in affected tissues remains limited [46]. Hence, it is imperative to conduct further research to uncover the functional implications of these genetic variants in intestinal inflammation and their interplay with nongenetic risk factors. Moreover, Genome-Wide Association Studies (GWAS) have identified more than 200 genomic regions associated with IBD; their ability to accurately pinpoint causal genes, their variations, and their functional consequences has been somewhat limited [64]. So, it is important to engage in additional research to pinpoint the precise genes and variants responsible for playing a role in intestinal inflammation and to acquire a deeper understanding of the mechanisms at their core. Recent studies have broadened our knowledge by revealing a growing spectrum of human monogenic diseases

that exhibit symptoms similar to intestinal inflammation observed in IBD [65]. However, additional research is required to identify the genetic irregularities responsible for these monogenic diseases and understand their fundamental mechanisms. Although current therapies for IBD can yield positive outcomes, they often have significant side effects and do not provide favorable results for all patients [66]. Therefore, it is crucial to conduct further research to create more targeted treatments that consider the unique environmental factors and genetics involved in intestinal inflammation.

The potential roles of non-coding genetic variations, gene-environment interactions, and gene-gene interactions in influencing susceptibility to IBD are subjects of investigation. GWAS have detected more than 200 genomic regions linked to IBD, and most of these regions are connected with DNA regulatory elements rather than protein-coding genes. This implies that non-coding genetic variants may substantially affect susceptibility to IBD. Recent studies have demonstrated that individual non-coding variants associated with IBD can influence DNA regulatory functions, potentially opening new therapeutic avenues to modulate transcription factor binding and enhancer activity [67]. IBD is a multifaceted condition characterized by numerous genetic and non-genetic risk factors that contribute to its development [17]. Although the exact mechanisms are not yet entirely understood, it is possible that gene-gene interactions could have an impact on IBD susceptibility. A recent study identified 63 out of 200 IBD risk loci as having enrichment in multigenic regulatory modules that include potential causative genes [68]. These findings indicate that gene-gene interactions might play a role in IBD susceptibility by potentially influencing the expression of multiple genes. Environmental factors, including diet, lifestyle, and microbiome, can also play a part in influence susceptibility to IBD [69]. The interplay between genes and the environment may contribute to IBD susceptibility by influencing gene expression during intestinal inflammation.

In individuals with a genetic predisposition, IBD is a complex condition stemming from a continuous and unregulated inflammatory reaction targeting the intestinal microbiome [70]. This suggests that genetic factors can interact with gut microbiota to influence susceptibility to IBD. An individual's genetic makeup and intestinal microbial community composition may contribute to their overall exposure to IBD [71]. Interactions between genes and gut microbiota can play a role in the initiation of IBD. Dysbiosis, which indicates an imbalance in the gut microbiota, could be a marker for interactions between the genome and microbiome [72].

Conclusion

In conclusion, IBD constitutes a complex and multifaceted category of gastrointestinal conditions, predominantly Crohn's disease and ulcerative colitis. These disorders arise from a blend of environmental factors and genetics, in which genetics substantially determines susceptibility to the disease. Genetic studies have identified numerous variants associated with IBD, including *NOD2, ATG16L1*, and *IL23R*. These variants affect various aspects of immune dysregulation, epithelial barrier function, and mucosal homeostasis, thereby contributing to the development and progression of IBD. However, it is essential to recognize that IBD is not solely a result of a single genetic mutation but rather a complex interplay of multiple genetic variations across different genes. Additionally, non-genetic factors, such as the gut microbiome and environmental triggers, play crucial roles in disease pathogenesis. Future research should focus on elucidating the functional consequences of these genetic variants within affected tissues, identifying causal genes and variants, exploring gene-gene interactions, and understanding the intricate interplay between the gut microbiome, genes, and environment in IBD susceptibility. This deeper understanding may pave the way for more precise diagnostic methods and personalized treatment strategies, ultimately improving the management and outcomes of individuals with IBD. Additionally, ongoing research may help to identify novel therapeutic targets and interventions that can better address the complexities of this chronic and debilitating condition.

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Инновации медицинского образования в хирургии желудочно-кишечного тракта с использованием интерактивных анатомических столов

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Внедрение цифровых технологий в процесс обучения студентов медицинских вузов является новой парадигмой медицинского образования, так как высокие критерии, предъявляемые к оценке качества подготовки врача, требуют использования современных технологий в преподавании фундаментальных дисциплин, и в частности анатомии. Благодаря симуляционным техникам создаются условия моделирования и интеграции обучающегося в условия, приближенные к реальным, что повышает интерес и вовлеченность студентов в образовательный процесс, и, как следствие, выявляется субъективное улучшение процесса усвоения материала. Классическое образование на анатомическом материале не может полностью обеспечить массовое обучение студентов, так как биологический материал не подлежит восстановлению и быстро повреждается — этих недостатков лишены цифровые трехмерные модели.

Цель исследования: оценить эффективность цифровой диссекции в образовательной траектории по клинической анатомии с использованием интерактивных анатомических столов и потребность обучающихся в активном применении указанных пособий.

Материалы и методы. Были сформированы 4 группы студентов: 3 группы изучали материал на различных анатомических столах, 1-я группа — по классической методике. Для контроля эффективности учебного процесса перед началом обучения во всех группах проведено входное тестирование. По окончании занятий по отдельным темам участники выполняли выходное тестирование, а также анкетирование.

Результаты. Анализ результатов обучения свидетельствует о значимо большем усвоении материала в группах, где во время занятий использовались трехмерные анатомические комплексы. Выявлено субъективное улучшение процесса усвоения знаний среди обучающихся за счет большей вовлеченности в образовательный процесс и повышения интереса к инновационным методикам проведения занятий.

Заключение. Результаты обучения будут изучаться в динамике, что позволит выявить различия в качестве формирования общепрофессиональных и профессиональных компетенций и послужит основой для принятия решения о повсеместном рутинном применении анатомических столов в образовательной практике.

Ключевые слова: медицинское обучение, цифровые технологии, трехмерная анатомия, трехмерное виртуальное моделирование, интерактивные анатомические столы, биологический материал, 3D-моделирование, контроль знаний

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