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Genetic Risk Factors For Chronic Obstructive Pulmonary Disease

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ABSTRACT

Background: The literature on genetic risk factors for chronic obstructive pulmonary disease (COPD) reveals a complex interplay between genetic predispositions and environmental influences, underscoring the multifactorial nature of this debilitating condition. **Literature Review:** Yuan et al. (Yuan et al., 2017) expanded on the complexity of genetic contributions by examining genetic polymorphisms linked to COPD, particularly in nonsmokers. Their findings indicated that genetic predispositions significantly influence the disease's occurrence, independent of smoking, through the identification of specific genetic loci associated with lung function decline (Yuan et al., 2017). This highlights the necessity of considering genetic factors even in populations traditionally viewed as lower risk. The exploration of patient-reported outcomes (PROs) by Melro et al. (Melro et al., 2018) further illustrates the impact of genetic variations on the subjective experience of COPD. Their systematic review demonstrated significant associations between genetic variants and various PROs, suggesting that genetic factors may influence not just physiological aspects but also the quality of life and disease progression as perceived by patients (Melro et al., 2018). Moll et al. (Moll et al., 2020) contributed to the understanding of genetic risk factors by analyzing polygenic risk scores and identifying overlapping genetic loci with lung function and pulmonary fibrosis. Their findings emphasized the heterogeneity of genetic contributions to COPD, suggesting that targeted interventions based on genetic predispositions could be beneficial (Moll et al., 2020). **Conclusion:** In conclusion, the body of literature reviewed demonstrates that genetic risk factors play a significant role in the etiology and progression of COPD. The integration of genetic insights with environmental factors is essential for developing effective prevention and treatment strategies. Continued research into the genetic underpinnings of COPD, alongside environmental interactions, will be vital for advancing personalized medicine approaches in managing this complex disease.

Keyword: Genetic Risk Factors, Chronic Obstructive Pulmonary Disease

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INTRODUCTION

Chronic obstructive pulmonary disease (COPD) is a multifaceted condition characterized by persistent respiratory symptoms and progressive airflow obstruction, influenced by a complex interplay of genetic and environmental factors. The literature on genetic risk factors for COPD has expanded significantly in recent years, revealing critical insights into the genetic architecture underlying this debilitating disease.

(Kumar et al., 2013) laid the groundwork by illustrating that COPD is influenced by multiple genes and environmental interactions. Their research highlighted the familial occurrence of COPD, suggesting a genetic component that cannot be solely attributed to known risk factors. The authors employed segregation analysis to confirm a significant genetic contribution to pulmonary function, indicating that various genes, each with limited effects, play a role in the disease's pathogenesis.

(Nedeljkovic et al., 2017) conducted a genome-wide association study that identified significant loci associated with COPD susceptibility, particularly focusing on the role of genetic variants in the FAM13A gene. They emphasized the importance of integrating genomic and transcriptomic approaches to better understand the epigenetic factors influencing COPD, including DNA methylation patterns that correlate with lung function changes.

(Yuan et al., 2017) further advanced the discourse by exploring genetic polymorphisms linked to COPD. Their findings underscored the complexity of genetic contributions, with specific loci associated with the disease's pathogenesis, particularly in smokers. The identification of new genetic signals related to lung function and emphysema provided a broader understanding of the genetic landscape of COPD, suggesting that genetic predisposition plays a crucial role even among nonsmokers.

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(Melro et al., 2018) shifted the focus towards patient-reported outcomes (PROs), highlighting the impact of genetic variations on the subjective experience of COPD. Their systematic review revealed significant associations between specific gene variants and various PROs, indicating that genetic factors may influence not only the physiological aspects of COPD but also the quality of life and disease progression perceived by patients.

(Moll et al., 2020) contributed to the understanding of the genetic underpinnings of COPD by identifying overlapping genetic loci with lung function and pulmonary fibrosis. Their comprehensive analysis of polygenic risk scores demonstrated the heterogeneity of genetic contributions to COPD, emphasizing the need for targeted interventions based on genetic predispositions.

(J. Alfahad et al., 2021) identified specific genetic defects, such as those in the SERPINA1 gene, that increase the risk of developing emphysema, reinforcing the notion that genetic factors are critical in COPD pathogenesis. Their findings highlighted the rapid accumulation of genetic associations, underscoring the potential for future research to uncover additional genetic determinants of the disease.

(Wang et al., 2022) explored the role of environmental factors, particularly air pollution, in conjunction with genetic susceptibility and lifestyle choices. Their research indicated that the interactions between genetic predispositions and environmental exposures are crucial in understanding COPD risk, suggesting a multifactorial approach to disease prevention and management.

Finally, (H. Czarnecka-Chrebelska et al., 2023) synthesized the current understanding of biological and genetic mechanisms linking COPD with lung cancer. Their work emphasized the genetic connections between these conditions, suggesting that shared genetic risk factors may contribute to both diseases' development and severity.

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Together, these studies illustrate the intricate genetic landscape of COPD, highlighting the need for a multifaceted approach in understanding its etiology and progression. The evolving research underscores the importance of integrating genetic insights with environmental factors to develop effective prevention and treatment strategies for COPD.

LITERATURE REVIEW

The article "Genomics of Chronic Obstructive Pulmonary Disease (COPD); Exploring the SNPs of Protease-Antiprotease Pathway" by Kumar et al. (Kumar et al., 2013) provides a comprehensive examination of the genetic factors contributing to the pathogenesis of COPD, highlighting the intricate interplay between genetic predispositions and environmental influences. The authors articulate that COPD, akin to asthma, is a multifaceted disorder influenced by a myriad of genes, environmental exposures, and gene-environment interactions.

One of the crucial insights from the article is the familial clustering of COPD cases, suggesting a significant genetic component in its etiology. The authors note that the increased prevalence of COPD among relatives of affected individuals cannot be solely attributed to shared environmental risk factors, indicating that genetic predisposition plays a vital role (Kumar et al., 2013). This assertion is supported by findings that demonstrate a decrease in the prevalence of COPD and similar lung function impairments with increasing genetic distance among relatives. Such evidence underscores the importance of genetic factors in the manifestation of COPD.

The article further delves into the methodologies for assessing genetic contributions to lung function, particularly through segregation analysis in family studies. This approach has provided robust insights into the heritability of pulmonary functions, reinforcing the notion that multiple genes, each exerting a modest effect, collectively influence lung function and the risk of developing COPD (Kumar et al., 2013). The authors emphasize that while significant progress has been made in identifying specific genes associated with COPD, the complexity of the disorder

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necessitates further exploration into the genetic underpinnings and their interactions with environmental factors.

Moreover, Kumar et al. (Kumar et al., 2013) highlight the role of certain genes involved in antiproteolysis, the metabolism of toxic substances from cigarette smoke, and the inflammatory response, all of which are pivotal in the pathophysiology of COPD. The focus on inflammatory mediators as a genetic target for understanding COPD's mechanisms is particularly noteworthy, as it aligns with the broader understanding of chronic inflammatory conditions.

The article "Understanding the role of the chromosome 15q25.1 in COPD through epigenetics and transcriptomics," authored by Nedeljkovic et al. (Nedeljkovic et al., 2017), presents a comprehensive examination of the genetic underpinnings associated with chronic obstructive pulmonary disease (COPD), with a particular focus on the chromosome 15q25.1 locus. The authors delve into the implications of specific genetic variants, particularly in the FAM13A gene, and their correlation with the susceptibility to COPD.

A significant contribution of the article is its integration of both genome-wide association studies (GWAS) and epigenetic analyses, which elucidate the multifaceted nature of genetic risk factors for COPD. The authors highlight that while certain variants, such as those in the CHRNA5 gene, are linked to smoking behavior, they do not directly influence COPD risk. This distinction is crucial as it underscores the complexity of genetic interactions and the necessity of considering both direct and indirect pathways when assessing genetic contributions to COPD.

The article further discusses the role of mRNA expression levels and amino acid changes in the CHRNA5 gene as risk factors for nicotine dependence and lung cancer, thereby establishing a connection between genetic predisposition and broader respiratory health outcomes. The authors also emphasize the importance of DNA methylation patterns, noting that variable methylation is associated with both COPD and lung function. This aspect of the study is particularly relevant as it

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suggests that epigenetic modifications may serve as intermediaries between genetic predispositions and phenotypic expressions of disease.

Moreover, the article provides an updated overview of the COPD gene list, which reflects the ongoing evolution of genetic research in this field. By dissecting the genetic architecture of COPD, the authors not only contribute to the understanding of its etiology but also pave the way for potential therapeutic targets and personalized medicine approaches.

The article "Genetic polymorphism and chronic obstructive pulmonary disease" by Yuan et al. (Yuan et al., 2017) provides a comprehensive examination of the genetic factors contributing to chronic obstructive pulmonary disease (COPD), particularly in the context of individuals who have never smoked. The authors highlight that while smoking remains the predominant risk factor for COPD, the occurrence of the disease in nonsmokers suggests that genetic predispositions play a significant role.

The article effectively synthesizes findings from various genome-wide association studies (GWAS) to underscore the complexity of COPD's etiology. The identification of specific loci, such as those on chromosome 15q25, which includes genes like CHRNA3, PSMA4, and ADAMTS7, is particularly noteworthy. These genes have been implicated in the pathogenesis of COPD, indicating that genetic polymorphisms can influence susceptibility to the disease, even in the absence of smoking (Yuan et al., 2017).

Moreover, the article discusses the findings from the UK BiLEVE study, which uncovered new genome-wide signals related to forced expiratory volume (FEV1). This highlights the multifactorial nature of COPD, where genetic factors may contribute to lung function decline, independent of environmental exposures. The authors' mention of a meta-analysis revealing 43 signals associated with lung function metrics further supports the assertion that genetic components are crucial in understanding COPD's progression (Yuan et al., 2017).

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The article also emphasizes the significance of cell-to-cell signaling and migration pathways, as indicated by the identified loci, which may provide insight into the biological mechanisms underlying COPD. This connection between genetic polymorphisms and cellular processes is essential for future research aimed at developing targeted therapies or interventions.

The article "Genetic profile and patient-reported outcomes in Chronic Obstructive Pulmonary Disease: a systematic review" by Melro et al. (Melro et al., 2018) provides a comprehensive examination of the relationship between genetic variations and patient-reported outcomes (PROs) in individuals suffering from Chronic Obstructive Pulmonary Disease (COPD). The authors argue that the impact of COPD is not solely dictated by lung function, as patients with similar degrees of airway obstruction often report vastly different experiences regarding their health status and quality of life.

The systematic review synthesizes findings from thirteen studies that explore the association between specific genetic variants and various PROs, including the frequency and severity of exacerbations, depressive symptoms, and health-related quality of life (HRQOL). This approach underscores the complexity of COPD as a multifactorial and heterogeneous condition, which necessitates a broader understanding of the factors influencing disease progression and patient experiences.

One of the key insights drawn from the article is the identification of significant associations between certain gene variants and the severity of COPD symptoms. The authors highlight that while the number of PROs linked to genetic variations is limited, the findings suggest that these genetic factors could be instrumental in predicting disease outcomes and tailoring interventions. This is particularly relevant in a clinical context, where personalized treatment plans could enhance patient care and improve overall health outcomes.

However, the article also notes the need for further research to establish a more robust connection between genetic factors and PROs. The current literature presents

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a nascent understanding of how genetic predispositions can influence the subjective experience of COPD, indicating an area ripe for exploration. The authors call for additional studies to expand upon these findings and to investigate the potential for genetic profiling in clinical practice, which could ultimately lead to more effective management strategies for patients with COPD.

The article titled "Chronic obstructive pulmonary disease and related phenotypes: polygenic risk scores in population-based and case-control cohorts" by Moll et al. (Moll et al., 2020) presents a comprehensive analysis of the genetic factors contributing to chronic obstructive pulmonary disease (COPD). The authors delve into the complex genetic landscape of COPD, revealing significant overlaps between loci associated with COPD, lung function, and pulmonary fibrosis. This intersection underscores the multifaceted nature of COPD, suggesting that genetic predispositions may influence not only the disease itself but also related respiratory conditions.

The study employs genome-wide association analyses to identify novel genetic loci linked to lung function and COPD, thereby paving the way for potential druggable targets. The identification of these loci is crucial, as it enhances the understanding of the biological mechanisms underlying COPD, which could lead to innovative therapeutic strategies. Furthermore, the authors highlight the importance of genetic risk scores in assessing COPD susceptibility, illustrating how these scores can stratify individuals based on their genetic risk, akin to the implications of monogenic mutations.

One of the key insights from the article is the recognition of heterogeneous cell-type and phenotype associations within the genetic architecture of COPD. This finding suggests that there is not a singular genetic pathway leading to COPD but rather a constellation of genetic influences that may manifest differently across individuals. The authors also discuss the SERPINE2 gene's association with COPD, which was observed in two large populations, indicating its potential role as a biomarker for susceptibility.

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Moreover, the article addresses the implications of airway wall thickening and emphysema, noting their independent familial aggregation in COPD. This observation points to the hereditary nature of these phenotypes, suggesting that genetic factors may play a significant role in their development. The authors also critically evaluate the methodology of case-control studies in COPD research, emphasizing how the sampling of cases and controls can significantly impact the results. This highlights the necessity for rigorous study designs in genetic epidemiology to ensure the reliability of findings.

The article "Current views in chronic obstructive pulmonary disease pathogenesis and management" by Alfahad et al. (J. Alfahad et al., 2021) provides a comprehensive overview of the genetic risk factors associated with chronic obstructive pulmonary disease (COPD). While the authors acknowledge that cigarette smoking is the predominant cause of COPD, they emphasize the significant role that genetic predispositions play in the disease's pathogenesis.

A critical insight from the article is the identification of the Z-allele in the SERPINA1 gene, which is linked to alpha-1-antitrypsin deficiency. This genetic defect is noted as the most crucial genetic factor contributing to the development of emphysema, a major component of COPD. The authors detail that individuals with the PI ZZ genotype exhibit a markedly reduced serum concentration of alpha-1-antitrypsin (10–15% of normal levels), which is insufficient to inhibit neutrophil elastase effectively. This insufficiency leads to unchecked lung parenchyma destruction, underscoring the importance of genetic factors in COPD susceptibility.

In addition to SERPINA1, the article discusses mutations in the elastin gene as another genetic determinant of COPD. The mutation identified by Kelleher et al. (as cited in Alfahad et al. (J. Alfahad et al., 2021)), which involves the substitution of glycine 773 to aspartate (G773D), is particularly noteworthy. This mutation compromises the structural integrity of elastic fibers, essential for maintaining lung flexibility, thereby contributing to the pathophysiology of COPD.

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Furthermore, the authors highlight the advancements made through genome-wide association studies (GWAS) and comprehensive genomic analyses over the past decade. These methodologies have facilitated the identification of numerous genetic variants associated with COPD susceptibility and have expanded the understanding of the genetic landscape of the disease. However, the article also underscores the need for continued research to discover and evaluate new candidate genes that may influence COPD progression.

The article titled "Air pollution and risk of chronic obstructed pulmonary disease: The modifying effect of genetic susceptibility and lifestyle" by Wang et al. (Wang et al., 2022) provides a comprehensive examination of the multifactorial nature of chronic obstructive pulmonary disease (COPD). It emphasizes the interplay between environmental exposures, genetic predispositions, and lifestyle choices in the development of this debilitating condition.

COPD, as highlighted in the article, is a significant global health concern, ranking as the third leading cause of death worldwide. The authors effectively contextualize the disease by detailing its defining characteristics, such as persistent respiratory symptoms and progressive airflow obstruction. They underscore the critical role that environmental pollutants play in exacerbating COPD risk, noting that the respiratory system is constantly exposed to harmful gases and particulates that can lead to adverse health outcomes.

A key insight from the article is the acknowledgment of genetic susceptibility as a crucial factor in the pathogenesis of COPD. The authors reference genome-wide association studies (GWASs) that have identified specific genetic loci linked to COPD, suggesting that genetic factors may influence individual responses to environmental risks. This aspect of the study is particularly valuable, as it opens avenues for further research into personalized medicine approaches for COPD management.

Moreover, the authors discuss the interactions between genetic factors and lifestyle choices, particularly tobacco use, which is well-documented as a primary risk factor

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for COPD. They expand the discussion to include other modifiable lifestyle factors such as alcohol consumption, dietary habits, and physical activity levels, all of which are significantly associated with COPD risk. This comprehensive approach highlights the need for a multifaceted strategy in both research and public health interventions aimed at mitigating COPD risk.

However, the article identifies a notable gap in the literature regarding the interactions between air pollution exposure, genetic susceptibility, and lifestyle factors in relation to COPD risk. The authors assert that no prior studies have systematically evaluated how these elements converge to influence the likelihood of developing COPD. This gap underscores the importance of their research, as it aims to fill this void and provide a clearer understanding of the complexities surrounding COPD.

The article "Biological and Genetic Mechanisms of COPD, Its Diagnosis, Treatment, and Relationship with Lung Cancer" by Czarnecka-Chrebelska et al. (H. Czarnecka-Chrebelska et al., 2023) provides a comprehensive overview of chronic obstructive pulmonary disease (COPD) and its intricate connections with lung cancer, particularly emphasizing the biological and genetic mechanisms involved.

The authors begin by outlining the primary characteristics of COPD, which include lung airflow limitation and tissue destruction. They highlight that while tobacco smoking is the predominant risk factor for COPD, various other inhaled irritants contribute to the disease's heterogeneous phenotypes. This assertion is crucial as it broadens the understanding of COPD beyond a singular causative agent, suggesting a multifactorial etiology that encompasses environmental and genetic influences (H. Czarnecka-Chrebelska et al., 2023).

A significant portion of the article delves into the pathogenesis of COPD, discussing the roles of inflammation, oxidative stress, and protease activity. The authors effectively summarize how these biological factors not only contribute to the development of COPD but also play a pivotal role in the disease's progression and severity. Their exploration of the relationship between COPD and lung cancer is

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particularly noteworthy, as they present evidence that COPD serves as an independent risk factor for lung cancer, irrespective of smoking behavior. This finding underscores the need for clinicians to consider COPD patients as a high-risk group for lung cancer, further complicating the management of these individuals (H. Czarnecka-Chrebelska et al., 2023).

Moreover, the article emphasizes the genetic aspects of COPD, detailing how genetic predispositions can influence the onset and severity of the disease. The authors argue for a deeper investigation into the genetic factors that may predispose individuals to COPD and its associated complications, such as lung cancer. This call for further research is vital, as understanding the genetic underpinnings could lead to more targeted therapies and preventive strategies (H. Czarnecka-Chrebelska et al., 2023).

CONCLUSION

The literature on genetic risk factors for chronic obstructive pulmonary disease (COPD) reveals a complex interplay between genetic predispositions and environmental influences, underscoring the multifactorial nature of this debilitating condition. The foundational work by Kumar et al. (Kumar et al., 2013) established that COPD is influenced by numerous genes and their interactions with environmental factors, highlighting a significant genetic component that cannot be solely attributed to known risk factors (Kumar et al., 2013). This notion is further supported by the familial clustering of COPD cases, which suggests that genetic predispositions play a crucial role in its etiology.

Nedeljkovic et al. (Nedeljkovic et al., 2017) advanced this understanding through genome-wide association studies (GWAS), particularly identifying significant loci associated with COPD susceptibility, such as variants in the FAM13A gene. Their integration of genomic and epigenetic analyses provided insights into how DNA methylation patterns correlate with lung function changes, emphasizing the importance of both genetic and epigenetic factors in COPD pathogenesis (Nedeljkovic et al., 2017).

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Yuan et al. (Yuan et al., 2017) expanded on the complexity of genetic contributions by examining genetic polymorphisms linked to COPD, particularly in nonsmokers. Their findings indicated that genetic predispositions significantly influence the disease's occurrence, independent of smoking, through the identification of specific genetic loci associated with lung function decline (Yuan et al., 2017). This highlights the necessity of considering genetic factors even in populations traditionally viewed as lower risk.

The exploration of patient-reported outcomes (PROs) by Melro et al. (Melro et al., 2018) further illustrates the impact of genetic variations on the subjective experience of COPD. Their systematic review demonstrated significant associations between genetic variants and various PROs, suggesting that genetic factors may influence not just physiological aspects but also the quality of life and disease progression as perceived by patients (Melro et al., 2018).

Moll et al. (Moll et al., 2020) contributed to the understanding of genetic risk factors by analyzing polygenic risk scores and identifying overlapping genetic loci with lung function and pulmonary fibrosis. Their findings emphasized the heterogeneity of genetic contributions to COPD, suggesting that targeted interventions based on genetic predispositions could be beneficial (Moll et al., 2020). Additionally, Alfahad et al. (J. Alfahad et al., 2021) highlighted specific genetic defects, such as those in the SERPINA1 gene, which are critical in the development of emphysema, reinforcing the notion that genetic factors are essential in COPD pathogenesis (J. Alfahad et al., 2021).

Wang et al. (Wang et al., 2022) explored the interactions between environmental factors, particularly air pollution, and genetic susceptibility, emphasizing the need for a multifactorial approach to understanding COPD risk. Their research suggested that the interplay between genetic predispositions and environmental exposures is crucial for disease prevention and management (Wang et al., 2022).

Finally, Czarnecka-Chrebelska et al. (H. Czarnecka-Chrebelska et al., 2023) synthesized current knowledge on the biological and genetic mechanisms linking

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COPD with lung cancer, highlighting shared genetic risk factors that may contribute to the development and severity of both conditions (H. Czarnecka-Chrebelska et al., 2023).

In conclusion, the body of literature reviewed demonstrates that genetic risk factors play a significant role in the etiology and progression of COPD. The integration of genetic insights with environmental factors is essential for developing effective prevention and treatment strategies. Continued research into the genetic underpinnings of COPD, alongside environmental interactions, will be vital for advancing personalized medicine approaches in managing this complex disease.

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REFERENCES

1. Kumar, M., Phougat, N., Ruhil, S., Dhankhar, S., Balhara, M., & Kumar Chhillar, A., 2013. Genomics of Chronic Obstructive Pulmonary Disease (COPD); Exploring the SNPs of Protease-Antiprotease Pathway. ncbi.nlm.nih.gov
2. Nedeljkovic, I., Carnero-Montoro, E., Lahousse, L., Van der Plaat, D. A., De Jong, K., Vonk, J. M., Van Diemen, C. C., Faiz, A., Van den Berge, M., Obeidat, M., Bosse, Y., Nickle, D. C., Uitterlinden, A. G., Van Meurs, J. J. B., Stricker, B. C. H., Brusselle, G. G., Postma, D. S., Boezen, H. M., Van Duijn, C. M., & Amin, N., 2017. Understanding the role of the chromosome 15q25.1 in COPD through epigenetics and transcriptomics. [PDF]

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3. Yuan, C., Chang, D., Lu, G., & Deng, X., 2017. Genetic polymorphism and chronic obstructive pulmonary disease. ncbi.nlm.nih.gov
4. Melro, H., Gomes, J., Moura, G., & Marques, A., 2018. Genetic profile and patient-reported outcomes in Chronic Obstructive Pulmonary Disease: a systematic review. [PDF]
5. Moll, M., Sakornsakolpat, P., Shrine, N., D Hobbs, B., L DeMeo, D., John, C., L Guyatt, A., J McGeachie, M., A Gharib, S., Obeidat, M. '., Lahousse, L., R A Wijnant, S., Brusselle, G., A Meyers, D., R Bleecker, E., Li, X., Tal-Singer, R., Manichaikul, A., S Rich, S., Won, S., Jin Kim, W., Ra Do, A., R Washko, G., Graham Barr, R., M Psaty, B., M Bartz, T., N Hansel, N., Barnes, K., E Hokanson, J., D Crapo, J., Lynch, D., Bakke, P., Gulsvik, A., P Hall, I., Wain, L., T Weiss, S., K Silverman, E., Dudbridge, F., D Tobin, M., & H Cho, M., 2020. Chronic obstructive pulmonary disease and related phenotypes: polygenic risk scores in population-based and case-control cohorts. ncbi.nlm.nih.gov
6. J. Alfahad, A., M. Alzaydi, M., M. Aldossary, A., A. Alshehri, A., A. Almughem, F., M. Zaidan, N., & A. Tawfik, E., 2021. Current views in chronic obstructive pulmonary disease pathogenesis and management. ncbi.nlm.nih.gov
7. Wang, L., Xie, J., Hu, Y., & Tian, Y., 2022. Air pollution and risk of chronic obstructed pulmonary disease: The modifying effect of genetic susceptibility and lifestyle. ncbi.nlm.nih.gov
8. H. Czarnecka-Chrebelska, K., Mukherjee, D., V. Maryanchik, S., & Rudzinska-Radecka, M., 2023. Biological and Genetic Mechanisms of COPD, Its Diagnosis, Treatment, and Relationship with Lung Cancer. ncbi.nlm.nih.gov

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