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Gene Technology

Short Communication

Personalised Therapy for Hereditary Diseases of Surfactant Dysfunction

Esti Gurber*

Department of Pharmacology, Ben-Gurion University of the Negev, Beer Sheva, Israel

DESCRIPTION

Hereditary diseases of surfactant dysfunction are a group of rare genetic disorders that affect the production, composition, or function of pulmonary surfactant a vital substance that lines the lung alveoli and facilitates proper respiratory function. These disorders can lead to severe respiratory distress in newborns and infants, and in some cases, chronic respiratory problems in adults. Advances in genetic study have enabled a deeper understanding the causes of these diseases,for personalized therapies for the specific genetic mutations involved [1]. Pulmonary surfactant is a complex mixture of lipids and proteins produced by specialized cells in the lungs called type II pneumocytes. Its primary function is to reduce surface tension in the alveoli, preventing their collapse during exhalation and ensuring efficient gas exchange.

Hereditary diseases of surfactant dysfunction are caused by mutations in genes associated with surfactant production or function. Mutations in genes such as *SFTPB*, *SFTPC*, *ABCA3*, and *NKX2-1* can lead to altered surfactant composition or impaired processing, resulting in respiratory distress [2]. Newborns with hereditary surfactant disorders often present with severe respiratory distress shortly after birth, a condition known as Neonatal Respiratory Distress Syndrome (NRDS). In some cases, respiratory symptoms may persist into childhood or adulthood, leading to interstitial lung disease. The genetic heterogeneity of these disorders contributes to variations in clinical presentation and severity [3]. Different mutations can lead to distinct phenotypes, making a precise diagnosis for understanding the specific nature of the disease in an individual patient. Personalized therapy for hereditary surfactant dysfunction involves tailoring treatments based on the specific genetic mutations identified in an individual. Understanding the genotype allows for a more targeted and effective approach to managing the underlying cause of the disorder. For certain genetic mutations that result in the absence or dysfunction of specific surfactant proteins, gene replacement therapy holds promise. This approach involves introducing functional copies of the defective gene into the affected cells, restoring normal protein production and function.

Some genetic mutations may lead to abnormal protein processing or function. Personalized pharmacological interventions, such as small molecules or compounds designed to correct specific defects, can be explored based on the identified genetic abnormalities. In cases where the primary issue is insufficient surfactant production, personalized therapy may involve surfactant replacement [4]. While this is a more established approach for Neonatal Respiratory Distress Syndrome (NRDS), tailoring the composition of the surfactant to match the specific deficits identified through genetic testing can enhance its effectiveness. For severe cases with irreversible lung damage, lung transplantation may be considered. Personalized therapy in this context involves careful evaluation of the patient's overall health, matching potential donors, and addressing any specific challenges associated with the underlying genetic condition. Personalized therapy extends beyond direct interventions to include supportive care to the individual's needs [5]. This may involve respiratory therapies, nutritional support, and management of associated complications based on the specific manifestations of the disorder. Given the rarity and genetic complexity of hereditary surfactant dysfunction disorders, there may be limited treatment options available. Developing targeted therapies for specific genetic mutations requires extensive research and resources. Personalized therapy relies on early and accurate diagnosis through genetic testing. Timely identification of the specific genetic mutations allows for the implementation of targeted interventions during critical developmental stages [6]. As with any personalized medicine approach ethical considerations related to genetic testing, informed consent, and potential societal implications must be addressed. Open communication between healthcare providers, patients and families for navigating these considerations. Personalized therapy often involves long-term follow-up to monitor treatment efficacy, adjust interventions as needed, and address evolving health needs. An inclusive and personalized care plan should include regular assessments and adjustments based on the individual's response to treatment. Managing hereditary diseases of surfactant dysfunction requires a multidisciplinary approach [7].

Correspondence to: Esti Gurber, Department of Pharmacology, Ben-Gurion University of the Negev, Beer Sheva, Israel, E-mail: estigur@gmail.com

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Collaborative care teams, including pulmonologists, geneticists, neonatologists, and other specialists, play a vital role in developing and implementing personalized therapy plans. Ongoing advancements in gene editing technologies, such as *CRISPR-Cas9*, hold promise for more precise and targeted interventions [8]. These technologies may enable the correction of specific genetic mutations associated with hereditary surfactant dysfunction. Continued into the genetic basis of surfactant dysfunction will likely lead to the identification of new genes and mutations [9]. This expanded genetic understanding will contribute to the development of novel therapies and further refine personalized treatment approaches. Participation in clinical trials and collaborative research efforts for advancing personalized therapy options. By engaging in these initiatives, patients and healthcare providers contribute to the collective knowledge base and facilitate the development of new and innovative treatments [10].

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