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Perspective

Therapeutic Strategies for Rare Diseases: Lysosome Dysfunction and Autophag

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DESCRIPTION

Rare genetic disorders affect only a tiny number of people, but because their causes are well recognized, accurate animal models and physiopathology research may be developed. Lysosomal storage disorders are a group of rare diseases characterized by a fundamental shift in lysosome activity. The frequent incidence of neurological symptoms in these disorders highlighted the significance of lysosomes in neurodegeneration. The same holds true for other subcategories of rare neurodegenerative illnesses. Lysosomes work with autophagosomes and endosomes to break down their contents *via* hydrolytic enzymes. Recently, it has become obvious that changes to the autophagy-lysosome pathway may play a role in neuronal death in a range of neurodegenerative diseases.

They use a number of rare neurodegenerative diseases to show that a variety of changes to the autophagy-lysosome pathway are associated with neuronal death. However, it is often unclear why changing this route can induce neurodegeneration. Macroautophagy, also known as autophagy in this context, is a conserved system that allows for the massive destruction of macromolecules or organelles by transporting them to lysosomes. A double-membrane compartment absorbs cellular substrates that need to be broken down, forming an autophagosome. Following its merger with the degradative organelles, lysosomes, hydrolytic enzymes break down their contents. This cellular process is important for the development of new therapeutics because it has been connected to a variety of neurodegenerative illnesses.

Although lysosomes are vital to this mechanism, it is unknown how autophagy is affected in many neurodegenerative illnesses. Lysosome function impairment is seen in a variety of uncommon neurodegenerative conditions with documented genetic changes. Developing remedies for each pathology is tough due to the variety of diseases and underlying mutations. However, understanding the genetic basis of these diseases provides an opportunity to investigate the components that influence the autophagy-lysosome pathway. In this sense, Lysosomal Storage Disorders (LSDs) are a group of disorders that predominantly disturb lysosomal function and are typically associated with impaired autophagy.

LSD patients typically exhibit neurological symptoms, highlighting the important role of lysosomes in sustaining neuronal survival. However, lysosomal dysfunction and diminished autophagy are also signs of other rare neurodegenerative diseases, either as a result of a basic failure or as a result of changes to other important pathways. They did not want to provide a comprehensive list of rare illnesses with lysosomal dysfunction in our work; instead, their selected a few cases to demonstrate the spectrum of altered functions and the multiple lysosome-autophagy pathway components that can be disrupted.

Lysosomes are critical components of the autophagy system because they degrade autophagic material after fusion with autophagosomes. Once again, LSD helped us comprehend the pathways that link lysosome malfunction to a faulty autophagy pathway. Most LSD produced by lysosomal dysfunction manifests as a decrease in autophagic flux, which assesses the rate at which the drug is eliminated *via* autophagy, as well as an increase in autophagosomes and the autophagic substrate p62. Similar changes are observed in the majority of rare neurodegenerative illnesses. Despite these similarities, autophagy abnormalities can be caused by failure at multiple phases of the process, such as the production of autophagosomes, the fusion of autophagosomes with lysosomes, the degradation of autophagosomes, or the recovery of the lysosome membrane following the completion of autophagy.

Rare neurodegenerative illnesses affect just a small number of people, making it challenging to develop a tailored treatment strategy for each condition. Instead, it would be more sensible to develop a small number of therapeutic techniques that can be utilized to treat certain subgroups of rare diseases with at least some similar physiopathology. Many rare neurodegenerative diseases are characterized by lysosome dysfunction, which frequently inhibits autophagy.

As previously noted, a few alterations, such as the accumulation of autophagosomes or autolysosomes, are observed in a large

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number of uncommon illnesses. The processes underlying these cellular dysfunctions, however, are frequently unknown. Lysosomes are also required for other functions besides autophagosome breakdown. Furthermore, they allow the breakdown of endocytosed macromolecules, and lysosome failure can disrupt the endocytosis mechanism.