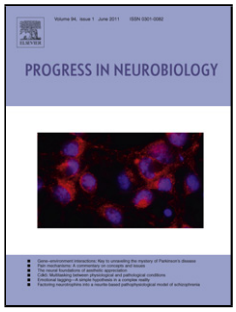

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Insights into the Pharmaceuticals and Mechanisms of Neurological Orphan Diseases: Current Status and Future Expectations

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Abstract

Several rare or orphan diseases have been characterized that singly affect low numbers of people, but cumulatively reach ~6% – 10% of the population in Europe and in the United States. Human genetics has shown to be broadly effective when evaluating subjacent genetic defects such as orphan genetic diseases, but on the other hand, a modest progress has been achieved toward comprehending the molecular pathologies and designing new therapies. Chemical genetics, placed at the interface of chemistry and genetics, could be employed to understand the molecular mechanisms of subjacent illnesses and for the discovery of new remediation processes. This review debates current progress in chemical genetics, and how a variety of compounds and reaction mechanisms can be used to study and ultimately treat rare genetic diseases. We focus here on a study involving Amyotrophic lateral sclerosis (ALS), Duchenne Muscular Dystrophy (DMD), Spinal muscular atrophy (SMA) and Familial Amyloid Polyneuropathy (FAP), approaching different treatment methods and the reaction mechanisms of several compounds, trying to elucidate new routes capable of assisting in the treatment profile.