عنوان مقاله:

Advances in personalized medicine in the ALS research

محل انتشار:

اولین کنگره پزشکی شخصی (سال: 1395)

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نویسنده:

Reza Rofougaran - *Förste forskningsassistent, Dept. of Phamacology and Clinical Neuroscience, Umeå University, Sweden*

خلاصه مقاله:

Amyotrophic lateral sclerosis (ALS) is a progressive neurodegenerative disorder that eventually leads to the death of motor neurons and fatal paralysis. Numerous different gene mutations have been found in familial cases of ALS, such as mutations in superoxide dismutase 1 (SOD1), TAR DNA-binding protein 43 (TDP-43), fused in sarcoma (FUS), C9ORF72, ubiquilin-2 (UBQLN2), optineurin (OPTN) and others. Importantly, the genetic and phenotypic heterogeneity of ALS leads to a variety of responses to similar treatment regimens. The application of personalized medicine in ALS first requires genetic screening among ALS patients. As better drugs become available, it will be important to take into account the genetic profile status of the patient to determine if individuals with certain mutations would respond better to particular treatments. The genetic screening has other advantage in which it is possible to study the connections between gene modifiers with age of onset and disease progression in ALS populations. The recent study that conducted at Umeå university showed, depending on the mutation, the progression rates, distribution, end-stage SOD1 aggregate levels, and histopathology were differed in ALS mice model. These results will be discussed. Next generation sequencing (NGS) technologies have emerged as a powerful tool for the genetic screening of causative mutations and subgrouping of ALS patients. In this section, I will present some recent results. In the presentation, in the context of personalized medicine, I will also discuss more about patient-derived fibroblast lines expressing different mutant SOD1 as a tool to study the role of misfolding SOD1 in the ALS pathogenesis. I consider that gene therapy has great potential for personalized medicine approaches in ALS, either by antisense oligonucleotide, small interference RNA or any other method such as antibodies targeting pathological proteins. These techniques have already been tested and appear to be effective in SOD1, TDP-43, C9ORF72 and FUS animal .models. I will review application of gene therapies relevant in personalized medicine approach in the ALS research

کلمات کلیدی:

personalized medicine

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