Computational and molecular analysis of RTN4 as novel therapeutic option for axonal regeneration

Sumaira Kanwal
COMSATS Institute of Information Technology, Pakistan

Demyelinating diseases, such as multiple sclerosis (MS), Charcot Marie Tooth (CMT) are accountable for a significant portion of the neurological disability burden worldwide, especially in young adults. Communication among the presynaptic terminus of a motor neuron and the postsynaptic membrane of a skeletal myofiber take place at the neuromuscular junction (NMJ). The fully-grown mammalian central nervous system (CNS) has a deprived aptitude to restore and return function after damage. Reticulon-4, also known as Neurite outgrowth inhibitor or Nogo, that has been identified as an inhibitor of neurite outgrowth specific to the central nervous system. A large number of genes have been associated with axonopathies and one of the emerging subgroups encodes membrane-shaping proteins with a central reticulon homology domain. This suggests that membrane-shaping disorders might be considered as a continuous disease-spectrum of the axon. The ligand shows the interaction with the residues at position of LEU133, lLE134, Pro135, LEU136, ILE155, ILE157, LEU158, ASP160, TYR161 These are the conserved residues and also found in the favored region. The analysis of RTN4 gene was performed on the DNA sample by amplifying and sequencing all the coding exons and their flanking intronic regions. However a missense mutation was found in exon 2 of RTN4 gene in the proband and later on whole family was analyzed. The pathogenicity of the mutation was checked by *in silico* analysis by using the SIFT and Polyphen. As it is well established, that RTN4 is involved in demyelinating diseases like MS so it can be directly involved in CMT 1 disease. On the other hand it can also be a genetic modifier through NgR-p75(NTR)-Mediated Signaling.

Speaker Biography

Sumaira Kanwal is working as Assistant Professor at Department of Bioscience in COMSATS institute of Information Technology, Sahiwal Pakistan. She was graduated from Kongju National University, South Korea as a human geneticist. Her basic interest in research is to find the genetic contribution primarily in Neuromuscular disorders, Neurological disorders and epigenetic of complex phenotypes of the epilepsy Genetics. Recently she did her post-doctoral research on the influence of micro RNAs on clinic-genomics of CMT1A. She has numerous international Publications on diverse subjects including Suicide, Polio eradication and role of Mitofusion2 in the development of diversity of phenotypes. She has been an invited speaker at various prestigious forums including International conference of the Genetics Society of Korea, 2016.

e: sumaira.kanwal@ciitsahiwal.edu.pk

Notes: