

# Central Nervous System and Therapeutics

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## New Phenotypic and Genotype SOD1 Mutation in dominant familial Motor Neuron Disease: A case report of a family and Epidemiology of Motor Neuron Disease in North Malabar

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Anterior horn cell diseases are a group of pure motor disorders involving both upper and lower motor neurons. They are currently untreatable and carry variable course based on the phenotype. The nervous system is vulnerable for oxidative stress due to its high oxygen consumption, low antioxidants, poor regenerative capacity, and presence of metal ions. A 36 year old female had exertion induced cramps of her right lower limb, coldness in the right lower limb, progressive weakness, wasting, and fasciculation; became bilateral within 4 years. She had cyanosis, hypothermia, and decreased sweating of right leg with psoriasis. Her nerve conduction and electromyography studies were suggestive of anterior horn cell disease, which was supported by histopathology. She had severe reduction in total volume of sweat produced and prolonged sweat latency on the right sided limbs as assessed by Quantitative sudomotor axon reflex test. DNA testing showed SOD1 cytogenetic band exon

4 of the SOD1 gene. chr. 21:33039650c>c/T; c. 319c>c/T. We report a new phenotype in dominantly inherited amyotrophic lateral sclerosis, with asymmetrical vasomotor and sudomotor changes and psoriasis. We would also like to present the epidemiologic data regarding the various clinical phenotypes of motor neuron disease in our part of the world.

### Speaker Biography

Soumya CV has worked as Consultant Neurologist in AKG Memorial Cooperative hospital, Talap, Kannur for 6 years from January 2013 to December 2018 Attends to around 45 OP patients per day regularly, has more than 30 IP patients per week, has thrombolysed more than 150 acute stroke cases personally, has given botulinum toxin to around 30 patients Working as part - time consultant Neurologist in Dhanalakshmi Hospital, Kannur from March 2018 with regular OPDs twice weekly. Started working in ASTER MIMS Hospital Kannur from February 2019

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