

5th European Conference on

Clinical and Medical Case Reports

September 07-08, 2017 Paris, France



Milen Velinov

NYS Institute for Basic Research in Developmental Disabilities, USA

Genomic medicine empowers personalized patient care-an example of effective albuterol treatment for a patient with homozygous DOK7 mutation

A 15 years old female patient was referred for genetic evaluation because of dysarthria and muscle hypotonic. Her early motor development was delayed. She did not start walking until 2.5 years. Reportedly her early speech development was not delayed. However at her first visit with us she was noted to have dysarthria. She was also reported to have academic difficulties, possibly associated with her dysarthria. This patient had prominent muscle weakness. She had great difficulty getting upright from squatting position. She could not run and sometimes had impaired balance. The patient's parents were first cousins. Screening laboratory tests and molecular testing for Friedreich ataxia were normal. Whole exome sequencing was done because no specific etiology was apparent. The patient tested positive for homozygous pathogenic sequence change in gene DOK7 (c.1124_1127dupTGCC). Homozygous mutations in this gene were previously associated with congenital myastenic syndrome type 10 (CMS10). Previously reported Albuterol treatment in 15 patients with DOK7 mutations led to improvement in these patients' muscle strength. Treatment of our patient was initiated with 4 mg P.O. Albuterol daily. She initially developed borderline increase in her blood pressure and the Albuterol dose was decreased to 2mg alternated with 4 mg daily. At this regimen no adverse effects were observed. Monthly fatigue tests examining the muscle strength were conducted and a significant improvement of muscle strength of hands, abdominal muscles and legs was recorded. In addition the patient's parents reported improvement in her speech. The patient continues to receive Albuterol treatment. Personalized medicine is a relatively new developing medical approach that already has wide application in cancer care and clinical genetics. This new field was greatly empowered by the development of technologies for massive nucleic acid sequencing. Identifying specific somatic or constitutive mutations helps the physicians develop patient-specific treatment plans that demonstrate better efficiency.

Biography

Milen Velinov is an Associate Professor at Albert Einstein College of Medicine and Director of the Genetic Service at Bronx-Lebanon Hospital. He is also the Program Director of Comprehensive Genetic Services and Specialty Clinical Laboratories at The New York State Institute for Basic Research in Developmental Disabilities. His research interests include Neuronal Ceroid Lipofuscinoses as well as rare/unique clinical manifestations and diagnoses in clinical genetics

Milen.Velinov@opwdd.ny.gov

Notes: