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Diagnosis and management of neurogenetic disorders: A practical approach

William S Baek

Parkside Medical Group, USA

Since the beginning of the 21st century the field of Neurogenetics has exploded, generating novel concepts, unveiling mechanisms, and creating the basis for innovative molecule-targeted specific therapies for neurological disorders. Establishing a genetic diagnosis for any neurological condition is critical for understanding the natural course of the disease and managing accordingly; it shall no longer be viewed as medically unnecessary. This has created a paradigm shift towards reclassifying diseases based on the molecular features rather than signs and symptoms. Down syndrome, 22q11.2 deletion syndrome, Angelman syndrome, Prader Willi syndrome, Klinefelter syndrome, Turner syndrome, cri-du-chat (5p deletion), phenyl ketonuria, neurocutaneous disorders, Duchenne's muscular dystrophy, Friedreich's ataxia (1/50,000), myotonic dystrophy, Huntington's disease (1/10,000), and Charcot-Marie-Tooth disease (1/3000) are among the most common hereditary neurological disorders. I would like to present several genetically confirmed cases seen in our outpatient clinic, including practical management of these conditions. This consists of a myriad of cases I have personally diagnosed and treated in an omnibus fashion, such as Fragile X syndrome, horizontal gaze palsy with progressive sclerosis (HGPPS), Smith-Magenis syndrome (SMS), Huntington's disease, spinocerebellar ataxia (SCA), oculopharyngeal muscular dystrophy (OPMD) and fascioscapulohumeral muscular dystrophy (FSHMD) with review of the literature.

Biography

William S Baek is a triple board-certified neurologist. He graduated from Seoul National University College of Medicine in 1999 and completed his Neurology residency at the University of Chicago and a fellowship in Clinical Neurophysiology at UC San Diego in 2006. He completed an NIH Post-doctorate research fellowship at the Children's Hospital of Philadelphia. He is on the Editorial Board for the *Journal of Neurology and Neuroscience* and *JSM Alzheimer's Disease and Related Dementia*. He has over 25 publications, almost all as sole author. He is a certified medical interpreter for Korean and Spanish. He provides Clinical Neurophysiology services (EEG, EMG, EP) as well as Botox injections. His research interests are Alzheimer's disease, Parkinson's disease, stroke, epilepsy, multiple sclerosis, ALS, Neuromuscular disorders, hereditary ataxias, migraine, autism, ADHD, Clinical Neurophysiology, Neurogenetics, language-concordance, ethics and professionalism.

william_s_baek@hotmail.com

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