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Master Thesis Defense

Entitled

IDENTIFICATION OF INHERITED AND DE NOVO EXOMIC VARIATIONS IN AN EMIRATI FAMILY WITH NEURODEVELOPMENTAL DISORDERS

by

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Abstract

Neurodevelopmental disorders are a heterogenous group of disorders that affect children at any point of development and lead to mental and motor function deficits. Often, the underlying cause could be genetic and inherited. This study investigated possible genetic variations that could have led to neurological abnormalities and other co-morbidities in an Emirati family. Whole exome sequencing was used to sequence the protein-coding regions of the genome to identify potential *de novo* and inherited variants that are associated with neurodevelopmental disorders in this family. A small set of variants, both *de novo* and inherited, were identified in high-risk genes associated with autism spectrum disorders and epilepsy.

Keywords: Neurodevelopmental disorders, autism spectrum disorder, epilepsy, SNPs, whole exome sequencing