

Patients clinical registry and epidemiology study of tuberous sclerosis in Qatar

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This proposal is looking to build patient's clinical registry for patients with tuberous sclerosis (TSC) and addressing the epidemiology of tuberous sclerosis in Qatar. TSC is a rare genetic disorder affecting multiple organ systems by the growth of non-malignant hematomas in brain, kidney, heart, liver, lungs or skin. TSC mainly results from mutations in either the TSC1 gene or TSC2 gene, which code proteins (Hamartin and Tuberin, respectively). The role of proteins is to regulate cell proliferation and differentiation. Losing the regulation will lead to abnormal development and generations of cells. TSC patients presented with diverse clinical phenotypes such as seizures, cognitive disabilities, behavioral issues, skin and eye abnormalities, kidney, heart and lungs problems. In addition, the

severity and complications of TSC have a wide spectrum that varies between patients, depending on the tumor's location and its size. Some patients may develop minimal effects such as skin changes and others may develop profound symptoms such as mental retardation. Patient management starts at birth and continued all through his life. Multidisciplinary team care is needed that include genetics, neurology, cardiology, oncology, neurosurgery; radiology, psychiatry; pediatric, nephrology and pulmonary. The clinical protocols for TSC are developed to treat and prevent disease complication according to patient age and clinical phenotypes.

Problem: In the Gulf region, specifically, in Qatar, there is a gap of knowledge about the epidemiology of TSC among the population. There is no existing study looked at the disease features, severity and management of TSC among patients in Qatar.

Objectives: The main objectives of this project are, to fill up the gap of knowledge of TSC,

to establish clinical patients registry, develop specialize clinic for TSC patients involving multidisciplinary teams and to establish stander protocols in the country. Hence, study the epidemiology of TSC in Qatar, improve the quality of life for patients and their families, improve the outcome and to obtain further research in the future.

Materials and Method: Data will be collected from medical records at Hamad Medical Corporation and Sidra medical clinical sites.

Biography

Munira Ali Aden has completed Bachelor degree in Nursing from University of Calgary in Qatar and pursuing her master degree in Genomics and Precision medicine at Hamad Bin Khalifa University- Qatar. Currently, Munira is working as a clinical nurse in neurology department at Sidra Medicine. Munira has created iCommunicate which uses cards with Arabic and English words written on them to help patients communicate across language barriers. Her Project was one of the winners in The World Innovation Summit for Health (WISH) and chosen to participate in WISH 2018 as part of Young Innovators Program.

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