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Preventive Measures to Reduce Spinal Muscular Atrophy Burden in regions with High Rate of Consanguinity

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Spinal Muscular Atrophy (SMA) is a rare group of autosomal recessive genetic disorders that cause progressive muscle weakness caused by disruptions in the SMN1 and 2 genes. Internationally, the prevalence of SMA is estimated at 1-2 per 100,000 persons, while incidence is estimated at 1 in 10,000 live births. In Saudi Arabia, the Arabian Gulf region, and most of the Arab countries, there is a high rate of consanguinity, with an estimated rate of first cousin marriages of 25 -35%. The carrier frequency internationally is one in fifty persons with a range of (30 - 80). The rate of carrier frequency in Saudi Arabia based on a population-based study was estimated at 2.6%. This leads to higher prevalence and incidence rates of this disease. We developed a model to estimate the birth incidence of SMA in Saudi Arabia and estimate the disease prevalence based on survival rate. The impact of factoring in the high rate of consanguinity and the carrier rate, our calculations have estimated a birth incidence of SMA of around 38 per 10,000 live births in the KSA, a rate much higher than the ones reported globally. The increased incidence and prevalence of this rare disease entail a major burden to the healthcare system in the country, which is majorly funded by governmental entities (namely the Saudi Ministry of Health (MOH)). SMA carriage screening is currently not part of pre marriage screening in the KSA. To decrease the rates of this rare inherited disease, we propose mandating pre-marriage screening for SMA carriage among Saudi Arabian citizens who are planning to get married. Screening would start with the male partner and in case of positivity, the female partner would be tested for gene carriage. Such a preventive measure will help mitigate the risk of increasing numbers of SMA patients in the country thus decreasing the burden of this disease. Pre marriage screening has a major regulatory and ethical impact which need be thoroughly studied before full consideration of this important initiative.

Keywords: Spinal Muscular Atrophy, rare disease, gene, healthcare, pre marriage screening

Biography

Prof. Mohammed AlJumah, Neurologist with wide clinical experience in neuromuscular diseases, neurogenetics, and neuroimmunology. He has contributed to the development of multiple health strategies and initiatives at a national and regional level.

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