Identification and Mapping of Locus on the Long Arm of Chromosome 4 that is Associated with Mental Retardation and Optic Atrophy in a Qatari Family with Undelineated Autosomal Recessive Disease

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Background: Autosomal recessive diseases are the single largest category of single-gene disorders among Arab population. A Qatari family includes 6 individuals from 3 related consanguineous sibships, with mental retardation. The clinical picture comprised significant mental retardation, retinal degeneration, optic nerve atrophy, and ataxic gait. They suffer also from oedemal puffiness of hands and feet. All parents and the unaffected family members are healthy.

Objective: Identification of undelineated autosomal recessive disorders among families in the Arab world, especially in the Gulf region.

Methods: Whole genome genotyping was done by (Illumina 300Kb SNPs), followed by homozygosity mapping and linkage analysis. Targeted resequencing of candidate genes was performed within the linked loci.

Results and conclusions: Homozygosity mapping revealed a 19.6 MB segment in the long arm of chromosome 4 flanked by rs4345237 (4q12) and rs422140 (4q13.3). This interval contains more than 100 genes, none of which has been implicated in any of the above mentioned phenotype so far. Candidate genes were selected and we are in the process of sequencing them. In addition, whole exome sequencing will be performed to expedite the mutation identification.

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Proportionate Mortality and Relative Risk from Motor Vehicle Crashes in Qatar: a Tool for Prioritizing Preventive Programs & Research

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Background: Motor vehicle crashes (MVCs) are a recognized public health problem in Qatar, where they are a leading cause of death. Given the diverse population in Qatar there is a need for evidence to identify high-risk populations bearing a disproportionate MVC mortality burden.

Objective: To identify the populations at the greatest risk for death from MVC's in Qatar, by measuring proportionate mortality (PMMVC) and relative risk (RRMVC).

Methods: An analysis of published mortality and population data was conducted to calculate for PMMVC and RRMVC and identify high-risk populations at a disproportionate risk for MVC mortality.

Results: One in seven (13.9%) deaths in Qatar is due to an MVC. Males are the victim in 90% of MVC deaths, with PMMVC > 30% from 5-29 years. One-half (51%) of all deaths in the 10-19 age group are due to MVC's, with the highest PMMVC for Qatari males (QM) aged 20-29 and for Non-Qatari males (NQM) aged 10-19. MVC's were the leading cause of death for QM's ages 5-39, Non-Qatari females (NQF's) ages 10-19, Qatari females (QF's) and NQM's ages 5-29. The highest RRMVC in the general population was for females ages 5-9 and males ages 10-19. The highest RRMVC is for QF's and QM's ages 20-29. NQF's also have a higher RRMVC than QM's for ages 10-19. The oldest and youngest populations had protective RRMVC's.

QM's have more than double the RRMVC, compared to the general population, from ages 5-39 and Qatari females (QF's) extend this risk till the age of 49. NQF's share this elevated risk profile [RRMVC>2] with QF's while NQM's are most at risk from 5-29 years.

Conclusion: MVC's are the leading killer of the largest segment of the population of Qatar, those ages 5-29. Young males [10-29 years] bear a disproportionate PMMVC but females have a higher RRMVC. An elevated RRMVC starts at 5 years and continues till 29 years. A commensurate response in the form of targeted interventions to reduce MVC deaths and research to identify and reduce risk factors amongst the populations with the highest MVC risk and burden should be prioritized.