AQUAPORIN LINKED MOLECULAR TOOLS FOR PERSONALIZED DIAGNOSTICS IN PREVENTING SUDDEN INFANT DEATH SYNDROME

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Abstract: Sudden Infant Death Syndrome (SIDS), a syndrome of concern in the State of Mississippi, is the unexpected death of a child under the age of one in which an autopsy shows no type of explanation of the cause of death. A variety of genetic factors have been proposed to predispose to SIDS. In a 2010 publication, Opdal SH and co-researchers identified single nucleotide polymorphism (SNP) linked to AQP4, a member of the aquaporin water transport protein, as a candidate SNP for genetic predisposition to SIDS (PubMed Identifier 20351659). The authors speculated that up-regulation of AQP4 molecules in the perivascular membranes in the brain, and thus edema and increased brain weight. Further, an association between the T allele and the CT/TT genotypes of Single Nucleotide Polymorphism (SNP) rs2075575 and cases of SIDS was observed. These genotypes were also found in SIDS cases with maternal nicotine use. We determined the frequency of these genotypes in populations as documented in dbSNP and found TT genotype was absent in a sub-Saharan Africa population. Additionally, published literature was searched for microRNA (miRNA) that could be investigated as regulators of AQP4 inhibitors or down-regulators, since they are proposed to reduce cytotoxic brain edema. We found that precursor miR-320a has been demonstrated as an inhibitor of AQP4. We propose that SNP rs2075575 and miR-320a be investigated as molecular tools for profiling genetic predisposition to SIDS. Our ongoing research involves use of visual analytics software to integrate and visualize the rs2075575 genotypes of study participants in the International HapMap project. The purpose is to gain deeper insights on the pedigree of individuals that have the CT/TT genotype.

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