Genetic and bioinformatic analysis of human aquaporin genes

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ABSTRACT

Aquaporins are transmembrane protein channels presented in all life forms from bacteria to human. Currently, there are 13 aquaporins in human which are differentially expressed in various tissues forming either aquaporin channels (transfer water) or aquaglyceroporin channels (transfer water and other solutes). Recently, mutations in human aquaporins have been reported to be associated with various diseases. In this study, full length cDNA sequences of human aquaporins were used to draw the phylogenetic relationship among them and to site the amino acid substitutions especially in the conserved NPA motifs and other conserved residues. The results indicated that, some aquaporins do not maintain the first conserved NPA motif, such as Aqua11 and Aqua12, or the two conserved NPA motifs like Aqua7. Other molecular features of human aquaporins were reported including the length of the N-terminus and the C-terminus and the spacer between the two conserved NPA motifs. The obtained phylogenetic trees using the nucleotide sequence of the coding regions or the amino acid sequences had different topologies. On both phylogenetic trees, the bacterial GlpF aquaglyceroporin did not come at the root of the tree, indicating that it came to existence after human aquaporins, contradicting the evolution theory of Darwin. The results of this study shed light on the possibility of using aquaporin SNPs as diagnostic tool for obesity.

Keywords: *Human aquaporins, SNPs, obesity, NPA motifs, phylogenetic tree.*

INTRODUCTION

Aquaporins are a family of small proteins (24 – 30 kDa) that form water channels in plasma membrane of living organisms presented in all forms of life from bacteria to human (Kruse et al., 2006 and Hub and Groot, 2008). They are subdivided into two main groups; transmembrane pure water channels (aquaporins) which conduct water but not solutes and aquaglyceroporins which transport water and solutes like glycerol (Borgnia et al., 1999; De Groot and Grubmuller, 2001; Fu et al, 2000 and Murata et al., 2000). Both types of channels are responsible for the passive transport of water and other molecules across biological membranes in all living organisms (Heller et al., 1980). Functionally, they are involved in many human diseases including cataracts (Shiels and Bassnett, 1996 and Berry et al, 2000), Sjogren's syndrome (Groneberg et al., 2002), brain edema after brain injury (Verkman et al., 2006). Genetic variants of members of aquaporin family were documented to be associated with obesity (Maeda et al., 2008) through the regulation of glycerol in adipocytes and liver. They are also involved in cell motility and wound healing (Verkman, 2009).