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Identification of disease causing mutations in hyperglycemic captive bred vervet monkeys (*Chlorocebus aethiops*)

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High levels of glycine were observed in the plasma (457-795 $\mu\text{mol/L}$, normal $<350 \mu\text{mol/L}$) and CSF (7.5-12.7 $\mu\text{mol/L}$, normal range 3-8) of a small percentage (8%) of captive bred vervet monkeys characterized with total cataract formation at Primate Unit and Delft Animal Centre. Although cataracts have been documented for a variety of primate species, hyperglycinemia as well as this rare and unusual association of conditions have not been reported in the literature before and clearly need elucidation. The purpose of this study was to investigate the disease causing genes in hyperglycinemic vervet monkeys. Eight animals were selected based on their cataract and hyperglycinemic status. The monkeys were assigned into a control and spontaneous (cataract/ hyperglycinemic) group. Gene expression and genotyping experiments were conducted using RNA and DNA samples extracted from blood. Three genes that are associated with nonketotic hyperglycinemia were prioritized, namely: Glycine dehydrogenase (*GLDC*), aminomethyltransferase (*AMT*) and Solute Carrier Family 6, Member 9 (*SLC6A9*). Genotyping analysis of the complete coding sequence of *GLDC*, *AMT* and *SLC6A9* revealed eight novel single base substitutions of which four were non-synonymous missense and four were silent nucleotide changes. For gene expression, *AMT* and *SLC6A9* were down-regulated in hyperglycinemic monkeys. Therefore, it is possible that *GLDC*, *AMT* and *SLC6A9* genes may be responsible for hyperglycinemia in captive bred vervet monkeys.

Biography

Zandisiwe Emilia Magwebu is currently a PhD student of South African Medical Research Council through the University of the Western Cape in South Africa. In 2014, she was a part of the Next Generation Scientist (NGS) group at Novartis Pharma, Basel, Switzerland.

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