Notes:

Meningococcal disease cases in Western Cape, South Africa screened for genetic deficiencies of complement components C5 and C6

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Black African (74) and Cape Colored (80) cases presenting with meningococcal disease (MD) were tested for genetic defects of terminal complement components C5 and C6. The C5 defects comprised known defects p.R1476X, p.Q19X and a third mutation, p.A252T, we believed to be pathogenic. It was originally detected in a black woman who is compound heterozygous (p.Q19X, p.A252T). She has very low C5 levels and had suffered recurrent MD. The C6 genetic defects investigated were the reported protein defects c.1086delA, c.1087delG, c.1403delC and c.2144. Results of genetic testing of 74 Black MD patients for p.A252T showed six children homozygous and C5 deficient. Their C5 levels had a range of the 0.1 to 0.3 µgm /ml (<4% of mean control). The protein is functionally active. Among the same group of black patients another 16 were C6 homozygous deficient (C6D). Among all 74 black patients, 22 (almost 30%) were C5D or C6D and required prophylaxis from further MD infections. Among 80 cape colored patients 8(10%) were C6D. No homozygous C5D colored patients were diagnosed. The reason that p.A252T is pathological and requires further study. Also we do not know how patients in other regions of Africa are affected. The Black Africans in the Cape Town area are mainly Xhosa and descendants of people from regions east and/or north of the Cape Town area. The problem may as well affect other African regions.

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Long-term potentiation requires unique postsynaptic SNARE fusion machinery

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Activity-dependent modifications in the neuronal synaptic connections which is also known as synaptic plasticity, underlie most of the fundamental adaptive features of the brain; one such example is strengthening of synaptic activity or long-term potentiation (LTP). Trafficking of the AMPA receptors to the post synaptic membrane of the excitatory synapses is critical during NMDA receptor mediated LTP, but the exact molecular machinery is unclear. This seminar will focus on the results that show vesicular membrane fusion is required for regulated AMPA receptor exocytosis during LTP and provide unique features of this particular SNARE-mediated fusion machinery. SNAP-47, a novel Q-SNARE protein in the post-synaptic fusion machinery, has been found to be important in LTP.

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