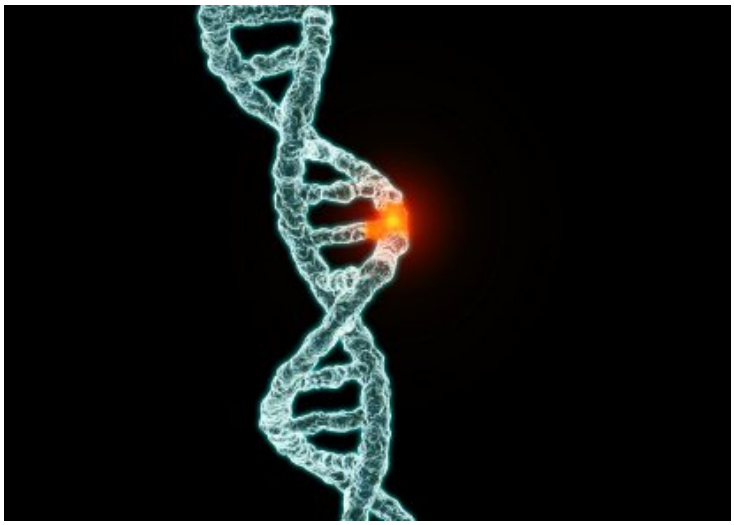
BioSpectrum the business of Bio & Health Sciences ASIA EDITION

Singapore scientists identify thick-skin-patch mutation

October 19, 2012 | News | By BioSpectrum Bureau

Singapore scientist identify mutation that causes skin hyperproliferation



Singapore: Scientists have identified a mutation in a gene that causes patches of very thick skin to appear on the palms and soles of affected people. These thick rough skin patches on hands and feet steadily increase in number as a person ages and often coalesce to form larger lesions. In severe cases, these lesions can be painful and debilitating.

The team of scientists from A*STAR's Institute of Medical Biology (IMB), in collaboration with hospitals and research centres from the UK, Japan and Tunisia, found that this skin disorder, called punctate palmoplantar keratoderma (punctate PPK), is caused by mutations in the AAGAB gene.

Punctate PPK is a rare subtype of palmoplantar keratoderma (PPK), which appears in subtly different forms and seems to have several possible causes. Several families in Singapore are afflicted by different types of PPKs and scientists at A*STAR have also been working with doctors at the National Skin Center to understand the different forms of this skin disorder.

The scientists analyzed DNA samples collected from 18 families from Scotland, Ireland, Japan and Tunisia who had punctate PPK. They showed that the AAGAB gene, which encodes the protein p34, was expressed in skin and had a role in the control of cell division.

The depletion in AAGAB led to a deficiency in p34, which resulted in increased cell proliferation in the outer layers of skin, the epidermis, because of an increased growth signal coming through the epidermal growth factor receptor (EGFR). The disruption of EGFR signalling is a feature of abnormal cell proliferation and the discovery suggests that PPK may be a benign form of hyperproliferation.