PXK gene: detailed annotation and new isoforms, associated with autoimmune disease Systemic Lupus Erythematosus (SLE)

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Systemic lupus erythematosus (SLE) is classified as a prototypic systemic autoimmune disease characterized by a very diverse range of clinical manifestations. Some patients show skin rashes but more than one-half of the SLE patients have more severe complications of the disease including glomerulonephritis, arthritis, central nervous system vasculitis, interstitial lung disease and stroke. SLE patients are suffering from disregulation of adaptive and innate immune systems. The principal immunological event in SLE is extended autoantibody production. The surplus of various immune complexes depositing in different organs results in inflammation and tissue damage.

The disease can occur at nearly any age. However, women in their reproductive ages are affected by the disease with the ratio of 9:1 to males. SLE is a complex disease which means that genetic factors and environmental factors such as sun exposure, certain drugs and viral infections contribute to the disease development.

Association of the Phox homology domain-containing serine/threonine protein kinase (PXK) gene to SLE has been reported recently by Genome Wide Association studies but the gene has not yet been well characterized. Our knowledge of functional annotation and causative variants of the gene is very insufficient.

In the current project; selective re-sequencing of some regions of the gene in a number of SLE patients and healthy individuals has been performed. A statistical genetic study on SNPs (single nucleotide polymorphisms) accomplished through genotyping study and statistical analysis.

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