GENETICS

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20.07.2009

Here is another reference to an earlier paper

Although a disease can be causally genetic, intensified mapping efforts have so far been unable to identify genes that account for more than a small fraction of the familial risk, perhaps because the responsible variation arises by somatic mutation (SM). SM explains the kind of epidemiological pattern seen in cancer, and might have a comparable role in many other diseases.

(I resisted to write about SM as did not want to be attracted by this Google search word). The prediction, however, seems to come true now by a Hum Mut paper

We found specific BAK1 single nucleotide polymorphism (SNP) containing alleles in both aneurysmic (31 cases) and healthy aortic tissue (5 cases) without seeing them in the matching blood samples. These same BAK1 SNPs have been reported, although rarely (average frequency <0.06%), in reference BAK1 DNA sequences. Based on this and other similar observations, we propose a novel hypothesis postulating that multiple variants of genes may preexist in minority forms within specific nondiseased tissues and be selected for, when intra- and/or extracellular conditions change.