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Germline variant in REXO2 is a novel candidate gene in familial pheochromocytoma

Eitan Friedman

The Susanne Levy Gertner Oncogenetics Unit, The Danek Gertner Institute of Human Genetics, Tel HaShomer, IsraelThe Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel.

Abstract

Pheochromocytoma (PCC) may be a rare, mostly benign tumor of the medulla . Hereditary PCC accounts for ~35% of cases and has been related to germline mutations in several cancer susceptibility genes (e.g., KIF1B, SDHB, VHL, SDHD, RET). We performed whole-exome sequencing during a family with four PCC-affected patients in two consecutive generations and identified a possible novel candidate pathogenic variant within the REXO2 gene that affects splicing (c.531-1G>T (NM 015523.3)), which co-segregated with the phenotype within the family. REXO2 encodes for RNA exonuclease 2 protein and localizes to 11q23, a chromosomal region displaying allelic imbalance in PCC. REXO2 protein has been related to DNA repair, replication and recombination processes and thus its inactivation may contribute to tumorigenesis. While the study suggests that this novel REXO2 gene variant underlies PCC during this family, additional functional studies are required so as to determine the putative role of the REXO2 gene in PCC predisposition. results show that ZXCII controls the sort I error rate well when there's no association. Furthermore, with regards to power, ZXCII is strong altogether of the situations considered and usually outperforms most of the prevailing methods within the presence of imprinting effects, especially under complete imprinting effects.

Biography

Prof. Eitan Friedman MD.PhD, Director and founder of the The Suzanne Levy-Gertner Oncogenetics Unit. MD graduate from the Sackler School of Medicine, TAU; PhD from the Karolinska Institutet, Sweden. The Oncogenetics Unit led by Prof. Friedman counsels and genetically tests individuals with a personal or family history of cancer. The main focus is breast and ovarian cancer but we offer services for all cancer types. Main research areas: inherited predisposition to cancer, population genetics, molecular mechanisms of Stress fracture development.