AB013. S013. Towards early detection of pancreatic cancer: applying NGS in the clinical setup

Merav Ben Yehoyada, Erez Scapa, Oren Shibolet, Erwin Santo, Guy Rosner

Tel-Aviv Sourasky Medical Center, Tel Aviv-Jaffa, Israel

Background: Pancreatic cancer (PC) is the third leading cause of cancer death in the US, with a lifetime risk of 1.5%. The 5-year survival rate is 7.2%, the poorest survival rate of any common malignancy. At early stages, surgical treatment is most beneficial, however, early diagnosis is rare, and most PC cases are confirmed as being locally invasive or metastatic. Inherited predisposition to pancreatic cancer is prevalent in 10% of PC cases. However, about 80% of PC cases with strong family history are still uncharacterized therefore making it hard to identify high-risk individuals. It was shown that PC development from a small precancerous lesion into a tumor is a relatively slow process, emphasizing the importance of screening in high-risk individuals. The concept of a dedicated high-risk pancreatic cancer clinic is relatively new and there are only a few such clinics worldwide. We established a high-risk pancreatic cancer clinic with the aims of (I) improving survival of individuals at high-risk for pancreatic cancer; (II) identifying genetic alterations and molecular pathways associated with familial pancreatic cancer risk.

Methods: We have recruited 135 high-risk PC individuals for genetic screening and clinical surveillance. Forty individuals fulfilled criteria for familial pancreatic cancer and 70% of the cohort have undergone genetic testing. We applied whole exom sequencing (WES) technique to screen families with high prevalence to PC for pathogenic germline mutations.

Results: Forty individuals (45% of those who were tested) were found to carry a pathogenic mutation. *BRCA2* mutation was found most frequently (20%), followed by *BRCA1* (14%), *PALB2*, *STK11* and *ATM* mutations. WES revealed a wide variety of genetic changes (*KLLN*, *HMMR*, *GATA5*, *MSR1* and *KDR* genes), that would not have been detected, if testing was limited to multi-gene panels.

Conclusions: Identifying high-risk PC individuals is crucial for surveillance and early detection that may lead to improved survival. WES is currently the test of choice for genetic evaluation of these high-risk individuals.

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