Research suggests a blood test to locate gene defects associated with cancer may not be far off

January 8 2014

Some surprising research findings from scientists at The University of Texas MD Anderson Cancer Center suggest it's possible a simple blood test could be developed to determine whether gene mutations associated with pancreatic cancer exist without the need of locating and testing tumor tissue. This appears possible following the discovery that tiny particles the size of viruses called 'exosomes,' which are shed by cancer cells into the blood, contain the entire genetic blueprint of cancer cells. By decoding this genomic data and looking for deletions and mutations associated with cancer, the research team believes this discovery could be translated into a test that helps physicians detect cancer and treat patients. The findings are based on research led by Raghu Kalluri, M.D., Ph.D., chairman and professor in MD Anderson's Department of Cancer Biology. The research results appear in the current online edition of the *Journal of Biological Chemistry*.

"At the present time, there is no single blood test that can screen for all cancer related DNA defects," said Kalluri. "In many cases, current protocols require a tumor sample to determine whether gene mutations and deletions exist and therefore determine whether the tumor itself is cancerous or benign. To procure tumor tissue, one needs to know that a tumor exists and if so, is it accessible for sample collection or removal? Finally, there are always risks and significant costs associated with surgical procedures to acquire tumor tissue."

Historically, researchers were aware these miniscule particles existed and that they carried nucleic acids and proteins. It was also believed that exosomes carried small portions of the person's DNA. However, upon further investigation, the MD Anderson research team was surprised to learn that the person's entire double-stranded genomic DNA spanning all chromosomes can be found in exosomes, including those mutated chromosomes that cause various cancers. Furthermore, Kalluri and colleagues discovered that DNA derived from exosomes carried the same cancer-related genetic mutations compared to the cancer cells taken from tumor.

"Because different forms of cancer are associated with different chromosomal mutations , we believe analysis of exosome DNA taken from blood samples may not only help determine the presence of a cancerous tumor somewhere in the body but also identify mutations without a need for tumor sample," added Kalluri. "We also believe this "fingerprint" will help lead us to the likely site of the tumor in the body. For instance, certain mutation spectrums would suggest pancreatic cancer or a brain-based tumor. While there is much more work to be conducted to develop such a test, having a tool such as this would increase our abilities to detect cancer in an earlier stage and therefore increase our chances of effective treatment."

"This seminal discovery paves the way for highly sensitive screening for driver mutations of cancer in the blood without the need for biopsy of tumor tissue and importantly, lays the foundation for a new method for the early detection of cancer when the chance for cure is greatest," said MD Anderson President Ronald A. DePinho, M.D.

Provided by University of Texas M. D. Anderson Cancer Center

Citation: Research suggests a blood test to locate gene defects associated with cancer may not be