Khan Inan Professor Truong BI-GY 7683

The types of mutations found within healthy cells are simply known as variants. Somatic mutations within cells can result in cancer but not always. Often when gene variants occur, they do not result in a self-proliferating mass of cells, which consequently does not lead to a tumor or cancer. The interesting thing is that these gene variants can occur and since they have little to no effect on the functionality of cells, a person might not ever even realize that they have happened. Gene. These random gene variations/mutations can occur due to a variety of factors but most commonly arise because of errors during DNA copying during cell division, or something like a virus infection that targets cells. Some of the types of gene variants that can occur are germline variants (which is a mutation that is consistent across all of an individual's cells) or a somatic variant which is a mutation found only in the newly mutated cells or cancer cells.

The main main difference between your average random mutation found in normal cells and cancer cells is that cancerous cells have mutated in such a way that is detrimental to the health of the person long term. For example, the lifespan of a cancer cell may be compromised and they may grow out of control or even spread to other parts of the body. On the other hand, mutations that have no effect on an organism or human may have had the mutation occur in a protein-coding region but does not affect the function of the protein that is created. If you were to look within a tumor from an organism then you would find that cells that should otherwise be resting and functioning normally are instead dividing out of control, and this in turn takes vital nutrients away from other cells and parts of the body (a lot like a parasitic relationship.)

To distinguish between healthy and cancerous cells using sequencing and statistical methods there are a few possible approaches. One approach is to look at the chromosomal differences between a normal and abnormal cell, because a cancerous cell will have a different number of chromosomes. In order to differentiate between what's normal and what's not in the first place it get a little bit more computationally intensive. One of the more recent methods, known as CopyKAT, uses single-cell RNA-sequencing data and data analytics to distinguish between cancer cells and normal cells. How it does this is that it uses sequencing data, and searches for aneuploidy (or abnormal chromones) using computer algorithms. This computational method is cutting-edge because not only can it distinguish between normal cells and cancerous cells, it can also identify sub-populations of other cancer cells within tumor, all by calculating and analyzing changes within genes and their number differences.

Cancer is mainly described as a genetic disease because tumors and cancerous cells are a result of changes in the genes that control a cell's ability to grow and multiply. It is often confused because people understand that you inherit these genes from your parents and family, but that is not what is meant by "genetic" in this case. Genetic in this case simply means that a

cell's genes are affected. In the case where an individual inherits an increased risk of cancer

associated with specific genes, it is called hereditary cancer...