Somatic Cell Genetics Encyclopedia Article

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Somatic Cell Genetics

Eukaryotic cells may be classified broadly as either germ cells (sex cells, or gametes) or all the other body cells, known as somatic cells. Somatic **cell genetics**, strictly speaking, means genetic studies of cells other than germ cells, but it is usually used more restrictively, referring to mammalian cells in tissue culture.

Key characteristics of somatic cells are that they are **diploid**, and differentiated. Though all somatic cells from one individual are (in principle) genetically identical, they evolve different tissue-specific characteristics during the process of **differentiation**, through regulatory and epigenetic changes.

The opportunity to study cells *in vitro* (isolated from the whole organism; literally, in glass) creates several advantages: one can assay cells in test tubes in ways that would be inappropriate to undertake on an individual, the cellular environment can be defined and manipulated, and rare genotypes can be maintained for study beyond the lifespan of the individual.

Most differentiated cells do not grow readily in culture; those that do tend to lose differentiated properties, and only have a finite lifespan (though they may be frozen for longer-term storage). Those that have arisen from tumors, on the other hand, or that are experimentally transformed *in vitro* may become permanent lines with indefinite lifespan. The most common source of tissue for non-transformed lines is skin fibroblasts. Lymphocytes can be cultured (short-term) from peripheral blood following stimulation by phytohemagglutinin, still the method used in diagnostic cytogenetics. Permanent lymphoblast lines are created with Epstein-Barr **virus transformation**, providing infinite supplies of otherwise rare genetic materials.

Perhaps the greatest contribution of somatic cell genetics was in the first stages of mapping the human **genome**. It was found in the 1960s that hybrids of human and rodent cells could be cultured, but selectively retained rodent and discarded human chromosomes. This phenomenon was taken to advantage, through selective culture conditions and identification of human chromosomes retained by the hybrids, allowing assignment of specific **gene** functions to specific chromosomes, starting with thymidine kinase to human **chromosome** 17. These early markers were the anchors for all later gene mapping studies.

In its broader sense, somatic cell genetics also encompasses the study of **cancer** cells, since the process of malignant transformation is one in which the genetic makeup of the **tumor** is altered relative to the otherwise stable constitution of the cells of origin.