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## Discovery Of Mobile Genetic Elements

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### 1983 Nobel Prize Medicine

Barbara McClintock was awarded the Nobel Prize in Physiology of Medicine of 1983 for her discovery of mobile genetic elements. She discovered that genes could be unstable; that there are certain regions of our genome that are capable of switching positions; and that this gene movement occurs more frequently than the rate at which mutations in the genome occur.

McClintock's discoveries were not appreciated by the scientific community in the late 1930's to the early 1950's. The reason behind this was that her discoveries were far ahead than their current bio-molecular and genetic knowledge. Nevertheless, a great appreciation of her work began with the development of molecular biology only in the late 1960's.

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## Background

During the late 1940's and early 1950's, the genetic code and the structure of the DNA double

helix were not yet known. The only known fact during those times was that the DNA molecule stores the genetic information in its structure. Then McClintock pointed out that there exist mobile genetic elements in the genome of plants and probably in our own genome too. She suggested that certain parts of our DNA are capable of changing positions, thus, causing a change in the functions of our genes.

It was not surprising that a lot of geneticists were not prepared to accept this concept of "jumping genes." It took decades of developments in the fields of molecular biology and genetics before it became apparent that the concept of jumping genes, if not universal, was quite common. These developments sparked further research in this field which produced outstanding discoveries which added on what McClintock has already contributed.

## **The Winner**

Barbara McClintock was born on the 16th of June 1902 in Hartford, Connecticut. She received her B.S. in 1923, earned her M.A. in 1925 and her Ph.D. in 1927 all from Cornell University. She then became a graduate assistant in the Department of Botany from 1924 to 1927. In 1927, she was awarded the position of Instructor which she held until 1931. She was then awarded a National Research Council Fellowship in 1931 and spent two years as a fellow at the California Institute of Technology.

In 1933 McClintock received another fellowship this time from Guggenheim. A year later, she went home to US and joined the Department of Plant Breeding at Cornell. In 1936, she accepted an assistant professorship in the Department of Botany at the University of Missouri. In 1941, she accepted a research offer from the Carnegie Institution of Washington in Cold Spring Harbor, New York. Here she was free to pursue any research that she thought of. McClintock remained at Cold Spring Harbor for the rest of her life. She died on September 2, 1992.

During McClintock's lifetime, she received numerous awards from different award-giving bodies. She received the Kimber Genetics Award, National Academy of Sciences in 1967, National Medal of Science in 1970, Lewis S. Rosenstiel Award for Distinguished Work in Basic Medical Research in 1978, The Louis and Bert Freedman Foundation Award for Research in Biochemistry also in 1978, Salute from the Genetics Society of America on August 18, 1980, Albert Lasker Basic Medical Research Award in 1981 and Louisa Gross Horwitz Prize for Biology or Biochemistry in 1982.

## **The Discovery**

Her researches started with an observation that in maize, kernels manifest different coloured patches. The prevailing answer to this observation was that certain chromosomes were more fragile than others, causing the genes in the fragile chromosomes to mutate more easily which causes unusual pigmentation. McClintock studied the structure of the chromosome of the kernels with the unusual pigmentation. She noted the structure, storage protein, starch content and the pigments in the individual chromosomes. From the ten pairs of chromosomes, she became particularly interested with chromosome pair nine.

It was indeed in chromosome pair nine where she found her first mobile genetic element which causes an interruption in the structure of the chromosome. She discovered that the

chromosome was divided into two by this mobile element that she called dissociation or Ds. Its transposition along chromosome nine caused an interruption to the usual sequence of genes in the chromosome causing some genes to be completely turned off. She also found that Ds need a trigger for it to be activated. This trigger was called activator or Ac. Together, the Ds and Ac were regarded by McClintock as a control mechanism of gene activity.

McClintock also found that these control elements are also present in different chromosomes but they act as normal genes. It is only after transposition that they cause inactivation of neighboring genes. She also found that these mobile genetic elements can be a part of regulatory control system in gene expression. Some elements act by programming neighboring genes to be activated or inactivated at a later time which may be several generations later.

## **Clinical Significance**

McClintock's discovery paved the way to the discovery of the role of mobile genetic elements in the spreading of resistance to antibiotics from resistant to sensitive strains of bacteria. The notion of transferable drug resistance in bacteria is a serious problem in the medical field because drug-resistant bacteria cause infections and disease that are more difficult to treat. This also entails that we need to develop alternative drugs that can treat the infections caused by the drug-resistant bacteria.

The discovery of mobile genetic elements also contributed to our knowledge of how antibodies are formed. Scientists have long been fascinated by the number of antibodies that our bodies can create considering that our genome has a very limited number of genes. The possibility of having jumping genes gave them the answer to this mystery. It is much like having a limited number of letters, but by having the ability to rearrange the letters, you can form almost an unlimited number of words.

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