The Philadelphia chromosome is a specific chromosomal abnormality that is commonly associated with chronic myeloid leukemia (CML) and a small percentage of acute lymphoblastic leukemia (ALL) cases. The Philadelphia chromosome results from a reciprocal translocation between chromosomes 9 and 22, where a piece of chromosome 9 breaks off and attaches to chromosome 22, and a piece of chromosome 22 breaks off and attaches to chromosome 9.

The resulting abnormal fusion gene, known as BCR-ABL1, encodes a protein with tyrosine kinase activity that promotes uncontrolled cell growth and proliferation. This leads to the development of leukemia.

The discovery of the Philadelphia chromosome and the BCR-ABL1 fusion gene has led to the development of targeted therapies, such as tyrosine kinase inhibitors (TKIs), which specifically target the activity of the BCR-ABL1 protein. These drugs have revolutionized the treatment of CML and have significantly improved patient outcomes.

The Philadelphia chromosome can be detected through cytogenetic testing, which involves analyzing the chromosomes of cancer cells. This can help to confirm a diagnosis of CML or ALL and can guide treatment decisions.