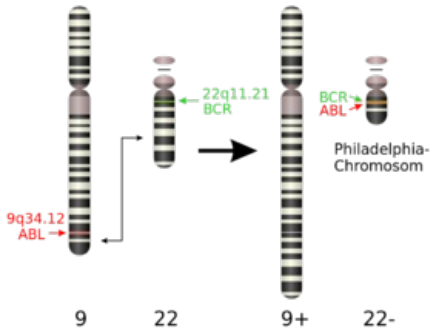
# Philadelphia chromosome



**The Philadelphia chromosome** is derived from chromosome 22. This chromosome is formed by a reciprocal translocation between chromosomes 9 and 22, in which the **ABL protooncogene is fused to the BCR gene** at the end of the long arm of chromosome 9. The BCR gene is located on the long arm of chromosome 22, and because it has a very strong promoter, this fusion results in the formation of an **active oncogene**. Due to the presence of a strong promoter, the protein resulting from the transcription of this **BCR-ABL fusion gene** is highly expressed, due to structural differences from the original kinases it does not respond to regulatory mechanisms (it is constantly active) and the cells are s stimulated to uncontrolled proliferation. The Philadelphia chromosome is very common in cancer patients with **chronic myeloid leukemia**.



Translocation scheme

In this case, it is an acquired chromosome aberration, ie a structural rearrangement of the chromosome, which occurs only during the patient's life. The Philadelphia chromosome is an example of a so-called **positional effect**. The ABL gene, which in its original position completely physiologically stimulates cells to proliferate, is brought close to a strong promoter as a result of translocation, thus significantly increasing the transcription of the translocated gene and then of the relevant protein. The resulting protein significantly stimulates cells to proliferate, and therefore translocation results in a **change of protooncogene to an active oncogene**.

This chromosomal aberration can be detected in patients by molecular cytogenetic examination, eg multicolour FISH (M-FISH), spectral karyotyping (SKY) or multicolour banding (M-BAND).

BCR-ABL protein can be inactivated by tyrosine kinase inhibitors such as Imatinib.

## Links

### Related articles

- Chromosomal aberrations in the etiology of neoplasms

### References

KOČÁREK, Eduard and Martin PÁNEK. Clinical cytogenetics I: introduction to clinical cytogenetics. 2nd edition. Prague: Karolinum, 2010. ISBN 978-80-246-1880-7.