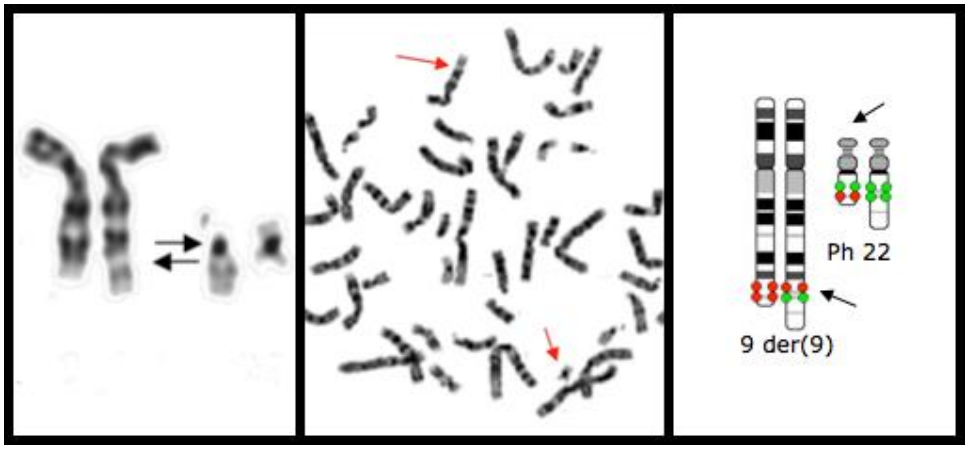
The Royal College of Pathologists
Pathology: the science behind the cure

Object 34: Philadelphia chromosome



What is it?
The Philadelphia chromosome is an abnormal chromosome 22, which forms following exchange of material with chromosome 9 in a process called translocation. This process occurs in a single bone marrow cell, which divides uncontrollably and passes the abnormal chromosome onto its daughter cells. This clonal expansion of the abnormal cell results in the person developing a type of leukaemia called chronic myeloid leukaemia (CML).

History
The Philadelphia chromosome was discovered in 1960 by American pathologists Peter Nowell and David Hungerford, and was the first chromosomal abnormality found to be linked to a specific form of malignancy. Until then, cancers were not thought to have a genetic cause. Nowell and Hungerford identified an unusually small chromosome 22 in patients with CML and named it the Philadelphia chromosome after the city where it was discovered.

Pathology
The abnormal chromosome results in the formation of an abnormal enzyme called a tyrosine kinase, which causes leukaemia cells to grow uncontrollably. The discovery of the Philadelphia chromosome led to major advances in the treatment of CML and the development of tyrosine kinase inhibitor drugs, such as imatinib mesylate (Glivec).

Find out more

Learn more about Howell and Hungerford's discovery and its significance on the Penn Medicine website.

Learn more about risk factors for CML on the Cancer Research UK website.