SUMMARY:

The Philadelphia negative chronic myeloproliferative neoplasms are chronic conditions, but in some cases may evolve into an "accelerated phase" or an acute leukemia. In particular in recent years several gene mutations and chromosomal alterations have been described, but neither the cause of chronic phase nor the molecular basis underlying their evolution have been completely clarified. In this study the researchers used high-resolution SNP analysis to study the cytogenetic alterations in these patients, which led to the identification of recurrent lesions: alterations of chromosome 1q and 9p were found to be associated with transformation into myelofibrosis post polycythemia vera/essential thrombocythemia or evolution in accelerated phase, while those of chromosomes 1q, 7q, 5q, 6p, 7p, 19q, 22q and 3q with transformation to acute leukemia. In some of these regions it has been possible to identify individual genes involved in the altered regions in all the patients, as FOXP1, TET2, IKZF1, CUX1, ETV6 and RUNX1, suggesting a possible role in the pathogenesis of these diseases. In conclusion, this study has allowed the identification of genetic lesions that can be implicated in the progression of these diseases.

To view the paper: http://www.ncbi.nlm.nih.gov/pubmed/21531982