Case report of isochromosome 17q in acute myeloid leukemia with myelodysplasia-related changes after treatment with a hypomethylating agent

J.C. Sousa1, R.T. Germano2, C.C.M. Castro3, S.M.M. Magalhaes1 and R.F. Pinheiro1,3

1Programa de Pós-Graduação em Ciências Médicas, Departamento de Medicina, Universidade Federal do Ceará, Fortaleza, CE, Brasil
2Universidade Estadual do Ceará, Fortaleza, CE, Brasil
3Departamento de Patologia, Universidade Federal do Ceará, Fortaleza, CE, Brasil

Corresponding author: R.F. Pinheiro
E-mail: ronaldpinheiro@pq.cnpq.br

Received November 8, 2011
Accepted March 27, 2012
Published August 6, 2012
DOI http://dx.doi.org/10.4238/2012.August.6.8

ABSTRACT. Isochromosome 17q is a relatively common karyotypic abnormality in medulloblastoma, gastric, bladder, and breast cancers. In myeloid disorders, it is observed during disease progression and evolution to acute myeloid leukemia in Philadelphia-positive chronic myeloid leukemia. It has been reported in rare cases of myelodysplastic syndrome, with an incidence of 0.4-1.57%. Two new agents have been approved for treatment of myelodysplastic syndrome/chronic myelomonocytic leukemia. These are the hypomethylating agents, 5-azacytidine and decitabine, recommended by consensus guidelines for high-risk myelodysplastic syndrome patients not eligible for
hematopoietic stem cell transplantation. We present a case of chronic myelomonocytic leukemia with normal cytogenetics at diagnosis treated with decitabine (with good response); however, the patient evolved to acute myeloid leukemia with i(17q) shortly after suspending treatment. To the best of our knowledge, this is the first report of acute myeloid leukemia with myelodysplasia-related changes with i(17q) after the use of a hypomethylating agent.

**Key words:** Myelodysplastic syndromes; Isochromosome 17q; Hypomethylation therapy