A TRANSLOCATION t(4;13)(q21;q14) AS SINGLE CLONAL CHROMOSOMAL ABNORMALITY IN A PARATHYROID ADENOMA

Gabriella Sammarelli¹, Marco Zannoni², Sabrina Bonomini¹, Roberto Delsignore¹, Vittorio Rizzoli¹, Mario Sianesi², and Cecilia Caramatti¹

¹Department of Internal Medicine and Biochemical Sciences, ²Department of General Surgery and Organ Transplantation, University of Parma, Parma, Italy

Most of the information about the genetic composition of parathyroid tumors has been obtained by comparative genomic hybridization (CGH) and loss of heterozygosity (LOH) studies, whereas only few conventional cytogenetic investigation results are available. We have performed cytogenetic analysis of short-term cultures from 3 parathyroid adenoma tissue samples. Two cases showed a normal karyotype in all the metaphases obtained from independent primary cultures. In one case 5 metaphases (in a total of 25) from 2 independent cultures showed a nonrandom translocation t(4;13)(q21;q14), which was therefore accepted as clonal. To our knowledge this is the second clonal translocation described in this tumor type. Further conventional cytogenetic analysis of more parathyroid tumor specimens would be necessary to identify other specific abnormalities and the involved genes with a potential important role in the diagnosis, prognosis and pathogenesis of parathyroid tumors.

Key words: comparative genomic hybridization (CGH), loss of heterozygosity (LOH), primary hyperparathyroidism (pHPT).