NF2 expression levels of gastrointestinal stromal tumors: a quantitative real-time PCR study

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ABSTRACT

Gastrointestinal stromal tumors are the most common mesenchymal tumors of the gastrointestinal tract. Until today, there have been few markers specific for the tumor. This has complicated the differential diagnosis of the neoplasm from tumors of smooth muscle origin. Recently, the proto-oncogene c-kit has been shown to be a very relevant marker as it almost invariably is expressed in gastrointestinal stromal tumors. Radiation exposure, hormonal and genetic factors, particularly neurofibromatosis 2, have been implicated in their development and growth. GIST initiation, either in NF2-associated or in sporadic cases, is linked to inactivation of members of the proteins 4.1 superfamily. The majority of the mutations identified in the NF2 gene result in a truncated protein and are clinically associated with a severe phenotype. Occasionally, missense mutations associated with a mild phenotype may occur. We compared NF2 gene expression in 5 cases with gastrointestinal stromal tumors by quantitative real-time polymerase chain reaction analysis. NF2 gene mRNA expression was assessed in fresh tissue of stomach from 5 consecutive patients. We detected no alterations in NF2 gene expression in the quantitative analyses of the 5 tumors.

Key words: gastrointestinal stromal tumors, neurofibromatosis 2, quantitative real time polymerase chain reaction.

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