R337H mutation of the TP53 gene as a clinical marker in cancer patients: a systematic review of literature

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ABSTRACT. The germline R337H mutation of the TP53 gene has been associated with the development of many tumor types. This systematic review of literature investigated the association between the R337H mutation and the patients’ family history and its predictive and prognostic value in cancer. Data were collected from articles archived in the PubMed, LILACS, MEDLINE, IBECS, and SciELO databases. The systematic review of literature was performed on 12 selected articles, describing a total of 175,462 individuals tested for the R337H mutation, including 1548 individuals with cancer and 118 individuals with a family history of Li-Fraumeni and Li-Fraumeni-like syndrome. Eight studies showed an association between the mutation and a family history of cancer in 411 patients, including 390 cases of cancer among family members. Patients with the homozygous mutant genotype experienced cancer recurrence, progressive disease, secondary cancer, and a short survival rate. Heterozygous patients showed a better response to treatment and increased survival rates than did patients with
the homozygous mutant genotype from newborns to adult patients. In conclusion, the R337H mutation has significant predictive and prognostic value and is associated with tumorigenesis of the adrenal cortex.

**Key words:** Adrenocortical carcinoma; Founder effect; Prognosis; Missense mutation; p.R337H