

Table 1. Frequency of the replication error phenotype in human cancers

Tumor type	RER, %*(n)†	Study
Familial (HNPCC)		
Colon (adenocarcinoma)	79 (14)	Aaltonen <i>et al.</i> [17**]
	86 (29)	Aaltonen <i>et al.</i> [22*]
Colon (adenoma)	57 (14)	Aaltonen <i>et al.</i> [22*]
Endometrial	75 (4)	Risinger <i>et al.</i> [23]
Sporadic		
Colon (adenocarcinoma)	13 (46)	Aaltonen <i>et al.</i> [17**]
	16 (49)	Aaltonen <i>et al.</i> [22*]
	12 (137)	Ionov <i>et al.</i> [24**]
	28 (90)	Thibodeau <i>et al.</i> [25**]
	16 (241)	Lothe <i>et al.</i> [42]
	13 (137)	Kim <i>et al.</i> [26**]
Colon (adenomas)	3 (33)	Aaltonen <i>et al.</i> [22*]
Endometrial	17 (36)	Risinger <i>et al.</i> [23]
	23 (30)	Burks <i>et al.</i> [48]
	20 (45)	Duggan <i>et al.</i> [49]
Blast crisis CML	74 (19)	Wada <i>et al.</i> [55]
Pancreatic	67 (9)	Han <i>et al.</i> [50]
Skin (squamous cell)	50 (10)	Zaphiropoulos <i>et al.</i> [54]
Skin (sebaceous tumors)‡	46 (13)	Honchel <i>et al.</i> [53]
Lung (small cell) (non-small cell)	45 (33)	Merlo <i>et al.</i> [56]
	34 (38)	Shridhar <i>et al.</i> [46]
	2 (87)	Peltomäki <i>et al.</i> [47]
Stomach	39 (57)	Han <i>et al.</i> [50]
	18 (33)	Peltomäki <i>et al.</i> [47]
	31 (52)	Rhyu <i>et al.</i> [51]
Renal cell	25 (36)	Uchida <i>et al.</i> [52]
Esophageal (adenoma)	22 (36)	Meltzer <i>et al.</i> [57]
Esophageal (squamous)	2 (42)	Meltzer <i>et al.</i> [57]
Ovary	16 (19)	Han <i>et al.</i> [50]
Cervix	15 (13)	Han <i>et al.</i> [50]
Bladder	3 (200)	Gonzalez-Zulueta <i>et al.</i> [58]
	21 (61)	Orlow <i>et al.</i> [59]
Breast	0 (84)	Lothe <i>et al.</i> [42]
	4 (26)	Han <i>et al.</i> [50]
	11 (104)	Wooster <i>et al.</i> [60]
	20 (20)	Yee <i>et al.</i> [61]
Testis	0 (86)	Lothe <i>et al.</i> [42]
	18 (69)	Murty <i>et al.</i> [62]
Prostate	3 (40)	Schoenberg <i>et al.</i> [63]
Liver	3 (29)	Han <i>et al.</i> [50]

*Percentage of tumors that exhibit at least one shifted locus.

†Number of cases analyzed.

‡Muir-Torre syndrome.

CML—chronic myelogenous leukemia.

17. Aaltonen LA, Peltomäki P, Leach FS, Sistonen P, Pylkkänen L, Mecklin JP, Järvinen H, Powell SM, Jen J, Hamilton SR, Petersen GM, Kinzler KW, Vogelstein B, de la Chapelle A: **Clues to the pathogenesis of familial colorectal cancer.** *Science* 1993, 260:812–816.

One of the three original papers describing the RER phenotype of microsatellite instability in CRC. These investigators described the finding in both familial (HNPCC) and sporadic cases.

22. Aaltonen LA, Peltomäki P, Mecklin JP, Järvinen H, Jass JR, Green JS, Lynch HT, Watson P, Tallqvist G, Juhola M, Sistonen P, Hamilton SR, Kinzler KW, Vogelstein B, de la Chapelle A: **Replication errors in benign and malignant tumors from hereditary nonpolyposis colorectal cancer patients.** *Cancer Res* 1994, 54:1645–1648.

This review describes the incidence of RER in precursor polyp lesions.

23. Risinger JI, Berchuck A, Kohler MF, Watson P, Lynch HT, Boyd J: **Genetic instability of microsatellites in endometrial carcinoma.** *Cancer Res* 1993, 53:5100-5103.

24. Ionov Y, Peinado MA, Malkhosyan S, Shibata D, Perucho M: **Ubiquitous somatic mutations in simple repeated sequences reveal a new mechanism for colonic carcinogenesis.** *Nature* 1993, 363:558-561.

One of the three original papers describing the RER phenotype of microsatellite instability in CRC. These investigators described the finding in sporadic cases.

25. Thibodeau SN, Bren G, Schaid D: **Microsatellite instability in cancer of the proximal colon.** *Science* 1993, 260:816–819.

Describes the RER phenotype of microsatellite instability in CRC. This is one of the three original papers where the investigators described the finding in sporadic cases.

26. Kim H, Jen J, Vogelstein B, Hamilton SR: **Clinical and pathological characteristics of sporadic colorectal carcinomas with DNA replication errors in microsatellite sequences.** *Am J Pathol* 1994, 145:148–156.

This review describes the clinical and pathologic features of 137 sporadic tumors exhibiting microsatellite instability of patients with Duke's disease stages B and C.

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