T75 disease 2394 2396 LS T76 cohort-patient 2466 2474 probands T73 mutation 2189 2200 p.Lys618Ala T74 mutation 2331 2342 p.Lys618Ala T79 size 1626 1630 2.8% T77 cohort-patient 882 890 carriers size 1571 1576 2.41% T78 cohort-patient 2485 2494 T29 relatives cohort-patient 2100 2114 positive cases T28 T27 cohort-patient 1489 1500 individuals T26 cohort-patient 940 951 individuals T25 cohort-patient 2303 2311 patients cohort-patient 1345 1353 T24 patients T23 cohort-patient 990 998 patients T22 cohort-patient 324 332 patients T21 body-part 966 976 colorectal T72 Disorder 2118 2144 microsatellite instability size 2096 2099 two T71 T20 disease 219 226 cancers T70 cohort-patient 1548 1552 nine T62 mutation 1367 1378 p.Lys618Ala T64 size 1476 1488 Twenty-seven mutation 1527 1538 T65 p.Lys618Ala T66 size 1548 1552 nine size 1579 1584 T67 seven T68 size 1636 1638 11 T69 cohort-patient 1644 1652 controls T38 disease 1827 1829 LS T37 mutation 1748 1759 p.Lys618Ala T39 cohort-patient 1579 1584 seven T34 cohort-patient 1847 1853 family T33 size 2152 2157 11.8% cohort-patient 689 707 control population T36 T35 cohort-patient 1949 1955 family T30 size 1105 1110 Three T61 size 1342 1344 17 cohort-patient 1830 1838 families T32 body-part 1307 1317 T60 colorectal cohort-patient 1133 1141 families T31 T41\_2 Concepts\_Ideas 145 173 autosomal dominant inherited disease 869 876 tumours T6 T7 disease 1318 1324 tumour T49 mutation 755 766 p.Lys618Ala T4 gene 1999 2003 MSH6 T48 mutation 653 664 p.Lys618Ala T5 disease 268 275 tumours T8 disease 2176 2183 tumours

Т9 disease 966 983 colorectal cancer T43 body-part 246 257 endometrium T94 Physiology 2264 2295 functional inactivation of MLH1 T93 mutation 1789 1809 pathogenic mutations body-part 234 244 T42 colorectum T41 disease 135 137 LS T40 mutation 31 47 c.1852\_1853AA>GC T47 disease 564 566 LS T46 mutation 405 416 p.Lys618Ala mutation 387 403 T45 c.1852\_1853AA>GC T44 disease 321 323 LS T88 Concepts Ideas 1610 1620 hereditary T89 cohort-patient 2081 2089 families T84 Disorder 207 218 early onset cohort-patient 1024 1032 T85 families T86 Concepts\_Ideas 957 965 sporadic T50 Disorder 839 865 microsatellite instability T87 Concepts\_Ideas 1557 1565 sporadic T1 aene 51 55 MLH1 T9\_cancer disease 977 983 cancer gene 2291 2295 MLH1 T3 gene 433 437 T2 MLH1 T59 mutation 1217 1228 p.Lys618Ala T10 disease 985 988 CRC T11 disease 1566 1569 CRC disease 1621 1624 CRC T12 disease 2299 2302 T13 CRC disease 2462 2465 T14 CRC T52 size 935 939 1034 T81 size 1813 1816 two T15 disease 81 95 Lynch syndrome T80 size 1654 1659 2.68% T51 mutation 920 931 p.Lys618Ala T16 disease 119 133 Lynch syndrome T54 250 size 1000 1003 T17 cohort-patient 1004 1018 index subjects T53 size 953 956 373 T82 size 1843 1846 one T18 size 2146 2150 2/17 T19 disease 174 180 cancer T56 cohort-patient 1094 1102 controls T55 disease 1053 1055 LS T58 disease 1130 1132 LS T57 size 1090 1093 411 R42\_m has Arg1:T28 Arg2:T33 R29\_2 relatedTo Arg1:T8 Arg2:T73 R15\_1 has Arg1:T27 Arg2:T65 R50\_1 has Arg1:T94 Arg2:T13

R9\_1 has Arg1:T31 Arg2:T59 R32\_m relatedTo Arg1:T13 Arg2:T3 R43\_1 has Arg1:T41\_2 Arg2:T41 R47\_2 has Arg1:T72 Arg2:T28 R48\_1 relatedTo Arg1:T21 Arg2:T10 R0\_T94\_T3 has Arg1:T94 Arg2:T3 R49\_2 has Arg1:T23 Arg2:T51 R20\_m has Arg1:T27 Arg2:T64 R46\_2 has Arg1:T24 Arg2:T7 R21\_m has Arg1:T87 Arg2:T11 R25\_2 has Arg1:T32 Arg2:T37 R4\_m has Arg1:T41\_2 Arg2:T16 R18\_m has Arg1:T24 Arg2:T61 has Arg1:T28 Arg2:T8 R30\_m R19\_m has Arg1:T24 Arg2:T62 has Arg1:T17 Arg2:T54 R13\_m has Arg1:T72 Arg2:T8 R28\_m R43\_m has Arg1:T32 Arg2:T38 R12\_m has Arg1:T23 Arg2:T53 R5\_m has Arg1:T84 Arg2:T20 R6\_m relatedTo Arg1:T42 Arg2:T20 R31\_m has Arg1:T76 Arg2:T14 R40\_m has Arg1:T70 Arg2:T78 R11\_m has Arg1:T26 Arg2:T52 R17\_m relatedTo Arg1:T60 Arg2:T7 R1\_m has Arg1:T1 Arg2:T40 R41\_m has Arg1:T28 Arg2:T18 R14\_m has Arg1:T56 Arg2:T57 R34\_m has Arg1:T86 Arg2:T9 R16\_m has Arg1:T31 Arg2:T30 R9\_m has Arg1:T2 Arg2:T45 R33\_m has Arg1:T25 Arg2:T13 R24\_m has Arg1:T32 Arg2:T81 R10\_2 has Arg1:T50 Arg2:T6 R39\_1 has Arg1:T3 Arg2:T73 R36 m has Arg1:T70 Arg2:T66 R26\_m has Arg1:T34 Arg2:T82 R51\_1 has Arg1:T41\_2 Arg2:T41 R15\_m has Arg1:T31 Arg2:T58 R27\_m has Arg1:T28 Arg2:T71 R52\_1 has Arg1:T41\_2 Arg2:T16 R2\_2 relatedTo Arg1:T15 Arg2:T1 R44\_m has Arg1:T2 Arg2:T46 has Arg1:T22 Arg2:T44 R8\_m R45\_m has Arg1:T77 Arg2:T49 R48\_m has Arg1:T77 Arg2:T6 R7\_m relatedTo Arg1:T43 Arg2:T20 R6\_1 has Arg1:T23 Arg2:T9

R39\_m has Arg1:T39 Arg2:T12 has Arg1:T41\_2 Arg2:T19 R3\_2 R0\_T21\_T9\_cancer relatedTo Arg1:T21 Arg2:T9\_cancer R32\_1 has Arg1:T69 Arg2:T80 has Arg1:T70 Arg2:T11 R37\_m R38\_m has Arg1:T39 Arg2:T67 R23\_m has Arg1:T69 Arg2:T68 R31\_1 has Arg1:T39 Arg2:T79 R22\_m has Arg1:T88 Arg2:T12