

## Gene Section

### Short Communication

# P53 (protein 53 kDa)

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### Identity

**Other names:** TP53 (Tumour Protein 53)

**Location:** 17p13

### DNA/RNA

#### Description

The gene encompasses 20 kb of DNA; 11 exons (the first is non-coding).

#### Transcription

3.0 kb mRNA; 1179 bp open reading frame.

### Protein

#### Description

393 amino acids; 53 kDa phosphoprotein.

#### Function

Tumour suppressor gene; P53 is a transcriptional regulator acting as a "guardian of the genome"; in response to DNA damage, p53 is overexpressed and activates the transcription of genes such as p21 (implicated in cell-cycle arrest) and BAX (implicated in apoptosis); these activations allow either the cells to repair DNA damage before entering further in the cell cycle, or to be eliminated. In both cases, the consequence is to prevent propagation of cells containing genetic alterations.

### Homology

5 highly-conserved regions between species. The p53 protein contains a transactivation domain, a DNA-binding domain, nuclear localization signals and a tetramerization domain.

### Mutations

#### Germinal

In Li-Fraumeni syndrome, a dominantly inherited disease in which affected individuals are predisposed to develop sarcomas, osteosarcomas, leukemias and breast cancers at unusually early ages.

#### Somatic

P53 is mutated in about 50% of human cancers, and the non-mutated allele is generally lost; the frequency and the type of mutation may vary from one tumor type to another; in general, mutations are found in the central part (exons 4-8) of the p53 gene; these mutations are missense, non-sense, deletions, insertions or splicing mutations; there are some hot-spots for mutations at CpG dinucleotides at positions 175, 248, 273 and 282.

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