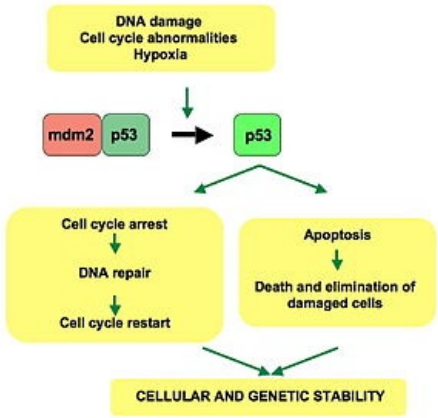
P53

The **TP53 gene** (Tumor protein p53) is one of the most important **tumor suppressor genes**. Its product, the p53 protein, acts as a transcription factor and as a **DNA damage sensor** in the cell. The p53 protein is nicknamed the "guardian of the genome" because of its key role in responding to genome damage. The TP53 gene, approximately 20 kb in size (11 exons), is located on the short arm of chromosome 17 (17p13.1). Homologous forms of the TP53 gene have been identified in most mammals, such as rats (chromosome 10), dogs (chromosome 5), and pigs (chromosome 12).

TP53 gene product - p53 protein

The p53 protein is a 53 kD nuclear protein (393 amino acids), a transcription factor, and a DNA damage sensor in the cell. In the physiological state, the p53 protein is inactive and is bound to the **MDM2** protein in the cytoplasm. When DNA is damaged, a signaling cascade is induced, which results in the phosphorylation of the MDM2 transporter and release of the p53 protein, thereby activating it. In the nucleus, it acts on the p21 gene, the product of which acts as an inhibitor of cyclin-dependent kinases. This **arrests the cell cycle** in the G1 phase, giving the cell time to repair. If the DNA repair is successful, the cell can continue progressing through the cell cycle. If the repair is unsuccessful, then the cell undergoes apoptosis (via the BAX protein signaling cascade).[1]



The role of p53 protein in various cellular processes

Li-Fraumeni syndrome

🔎 *For more information see Li-Fraumeni syndrome.*

The TP53 gene plays a key role in the pathogenesis of Li-Fraumeni syndrome,[2] a rare syndrome caused by a germline mutation in this gene. It manifests itself in very different ways, but it is especially associated with numerous soft tissue tumors (sarcomas, CNS tumors, carcinomas). The mode inheritance of the syndrome is autosomal dominant with incomplete penetration.

References

Related articles

- Li-Fraumeni syndrome
- Tumor suppressor genes

Citations

1. NOVOTNÁ, Božena a Jaroslav MAREŠ. *Vývojová biologie pro mediky.* 1. vydání. Praha : Karolinum, 2005. 99 s. ISBN 80-246-1023-X.
2. ŠÍPEK, Antonín. *Syndrom Li-Fraumeni* [online]. [cit. 2012-02-20]. <https://www.wikiskripta.eu/w/Syndrom_Li-Fraumeni>.

Used literature

- NOVOTNÁ, Božena a Jaroslav MAREŠ. *Vývojová biologie pro mediky.* 1. vydání. Praha : Karolinum, 2005. 99 s. ISBN 80-246-1023-X.