ERCC1

DNA excision repair protein ERCC-1 is a protein that in humans is encoded by the ERCC1 gene.

Together with ERCC4, ERCC1 forms the ERCC1-XPF enzyme complex that participates in DNA repair and DNA recombination.

Some DNA repair genes may be associated with pathogenesis and poor prognosis of astrocytoma [1)].

Targeting of DNA-repair genes such as Ercc1 could be used as an adjuvant chemosensitization treatment, similarly to Mgmt inhibition [2)].

The ERCC1-XPF nuclease is an essential activity in the pathway of DNA nucleotide excision repair (NER). The ERCC1-XPF nuclease also functions in pathways to repair double-strand breaks in DNA, and in the repair of “crosslink” damage that harmfully links the two DNA strands.

Cells with disabling mutations in ERCC1 are more sensitive than normal to particular DNA damaging agents, including ultraviolet (UV) radiation and to chemicals that cause crosslinking between DNA strands. Genetically engineered mice with disabling mutations in ERCC1 have defects in DNA repair, accompanied by metabolic stress-induced changes in physiology that result in premature aging.

Complete deletion of ERCC1 is incompatible with viability of mice, and no human individuals have been found with complete (homozygous) deletion of ERCC1. Rare individuals in the human population harbor inherited mutations that impair the function of ERCC1. When the normal genes are absent, these mutations can lead to human syndromes, including Cockayne syndrome (CS) and COFS.

ERCC1 and ERCC4 are the gene names assigned in mammalian genomes, including the human genome (Homo sapiens). Similar genes with similar functions are found in all eukaryotic organisms.

1)
Jiang Z, Hu J, Li X, Jiang Y, Zhou W, Lu D. Expression analyses of 27 DNA repair genes in astrocytoma by TaqMan low-density array. Neurosci Lett. 2006 Dec 1;409(2):112-7. Epub 2006 Oct 10. PubMed PMID: 17034947.
2)
Boccard SG, Marand SV, Geraci S, Pycroft L, Berger FR, Pelletier LA. Inhibition of DNA-repair genes Ercc1 and Mgmt enhances temozolomide efficacy in gliomas treatment: a pre-clinical study. Oncotarget. 2015 Aug 20. [Epub ahead of print] PubMed PMID: 26336131.