

ERCC8 (excision repair cross-complementing rodent repair deficiency, complementation group 8)

Identity

Note see also the paper on [Nucleotide Excision Repair](#)

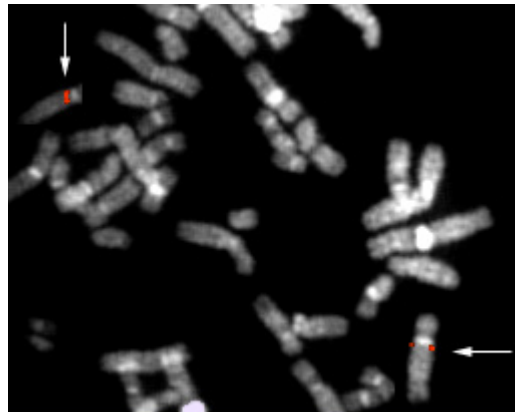
Other names **CSA (Cockayne syndrome A)**

CKN1

ERCC8

Hugo [ERCC8](#)

Location 5



bA489L13

[CSA \(5\)](#) - Courtesy Mariano Rocchi, [Resources for Molecular Cytogenetics](#).
Laboratories willing to validate the probes are welcome : contact rocchi@biologia.uniba.it

Note see also the paper on [Nucleotide Excision Repair](#)

DNA/RNA

Transcription 2011 b

Protein

Description 396 amino acids - 44 kDa

Function The Cockayne syndrome group A (CSA) gene encodes a WD repeat protein that interacts with the Cockayne syndrome group B ([CSB](#)) protein and a subunit of RNA polymerase II transcription factor TFIIH suggesting that the products of CSA and CSB genes are involved in transcription. The CSA defect leads to defective strand specific repair of

transcriptionally active genes.

Mutations

Germinal one base substitution

Implicated in

Entity Cockayne syndrome, CS group A

Disease The Cockayne syndrome A is characterized by sensitivity to sunlight, dwarfism, precociously senile appearance, pigmentary retinal degeneration, optic atrophy and deafness.

External links

Nomenclature

[Hugo](#)

[ERCC8](#)

[GDB](#)

[ERCC8](#)

[Entrez_Gene](#)

[ERCC8_1161](#) excision repair cross-complementing rodent repair deficiency, complementation group 8

Cards

[Atlas](#)

[CSAID301](#)

[GeneCards](#)

[ERCC8](#)

[Ensembl](#)

[ERCC8](#)

[Genatlas](#)

[ERCC8](#)

[GeneLynx](#)

[ERCC8](#)

[eGenome](#)

[ERCC8](#)

[euGene](#)

[1161](#)

Genomic and cartography

[GoldenPath](#)

[ERCC8 - 5 chr5:60205415-60276648 - 5q12.1](#) (hg18-Mar_2006)

[Ensembl](#)

[ERCC8 - 5q12.1 \[CytoView\]](#)

[NCBI](#)

[Genes Cyto](#) [Gene Seq](#) [Map View - NCBI]

[OMIM](#)

[Disease map \[OMIM\]](#)

[HomoloGene](#)

[ERCC8](#)

Gene and transcription

[Genbank](#)

[AK056931](#) [ENTREZ]

[Genbank](#)

[AL691658](#) [ENTREZ]

[Genbank](#)

[BC009793](#) [ENTREZ]

[Genbank](#)

[BT020021](#) [ENTREZ]

[Genbank](#)

[CR536563](#) [ENTREZ]

[RefSeq](#)

[NM_000082](#) [SRS] [NM_000082](#) [ENTREZ]

[RefSeq](#)

[NM_001007233](#) [SRS] [NM_001007233](#) [ENTREZ]

RefSeq	NM_001007234 [SRS] NM_001007234 [ENTREZ]
AceView	ERCC8 AceView - NCBI
TRASER	ERCC8 Traser - Stanford
Unigene	Hs.435237 [SRS] Hs.435237 [NCBI] HS435237 [spliceNest]
Protein : pattern, domain, 3D structure	
SwissProt	Q13216 [SRS] Q13216 [EXPASY] Q13216 [INTERPRO]
Prosite	PS00678 WD_REPEATS_1 [SRS] PS00678 WD_REPEATS_1 [Expasy]
Prosite	PS50082 WD_REPEATS_2 [SRS] PS50082 WD_REPEATS_2 [Expasy]
Prosite	PS50294 WD_REPEATS_REGION [SRS] PS50294 WD_REPEATS_REGION [Expasy]
Interpro	IPR001680 WD40 [SRS] IPR001680 WD40 [EBI]
CluSTr	Q13216
Pfam	PF00400 WD40 [SRS] PF00400 WD40 [Sanger] pfam00400 [NCBI-CDD]
Smart	SM00320 WD40 [EMBL]
Prodom	PD000018 WD40 [INRA-Toulouse]
Prodom	Q13216 ERCC8 HUMAN [Domain structure] Q13216 ERCC8 HUMAN [sequences sharing at least 1 domain]
Blocks	Q13216
HPRD	Q13216
Protein Interaction databases	
DIP	Q13216
IntAct	Q13216
Polymorphism : SNP, mutations, diseases	
OMIM	216400;609412 [map]
GENECLINICS	216400;609412
SNP	ERCC8 [dbSNP-NCBI]
SNP	NM_000082 [SNP-NCI]
SNP	NM_001007233 [SNP-NCI]
SNP	NM_001007234 [SNP-NCI]
SNP	ERCC8 [GeneSNPs - Utah] ERCC8 [HGBASE - SRS]
HAPMAP	ERCC8 [HAPMAP]
General knowledge	
Family Browser	ERCC8 [UCSC Family Browser]
SOURCE	NM_000082
SOURCE	NM_001007233
SOURCE	NM_001007234

SMD	Hs.435237
SAGE	Hs.435237
Amigo	nucleotide-excision repair complex
Amigo	protein binding
Amigo	nucleus
Amigo	transcription-coupled nucleotide-excision repair
Amigo	transcription
Amigo	regulation of transcription, DNA-dependent
Amigo	sensory perception of sound
Amigo	nuclear matrix
PubGene	ERCC8

Other databases

Other database [Cockayne syndrome](#)

Probes

[Probe](#) [Cancer Cytogenetics \(Bari\)](#)
[Probe](#) [ERCC8 Related clones \(RZPD - Berlin\)](#)

PubMed

[PubMed](#) [14 Pubmed reference\(s\) in LocusLink](#)

Bibliography

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Henning K. A., Li L., Iyer N., McDaniel L. D., Reagan M. S., Legerski R., Schultz R. A., Stefanini M., Lehmann A. R., Mayne L. V. and et al.
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Medline [Ozdirim](#)

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Medline [Stefanini](#)

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[BiblioGene - INIST](#)

Contributor(s)

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