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Gene Section
Mini Review

RECQL4 (RecQ protein-like 4)

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Identity

Other names: RecQ4; RTS; RecQ protein like 4; ATP-dependent DNA helicase Q4

HGNC (Hugo): RECQL4

Location: 8q24.3

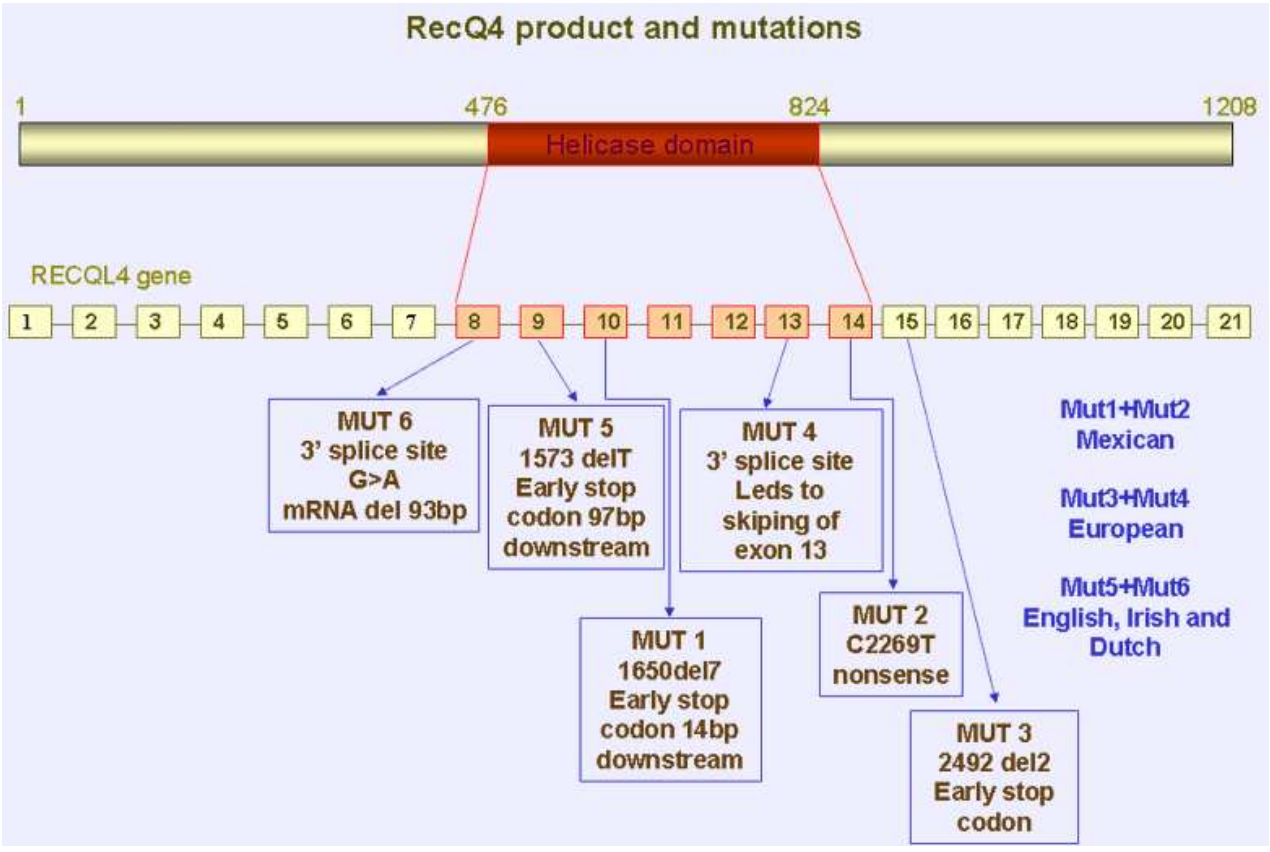
DNA/RNA

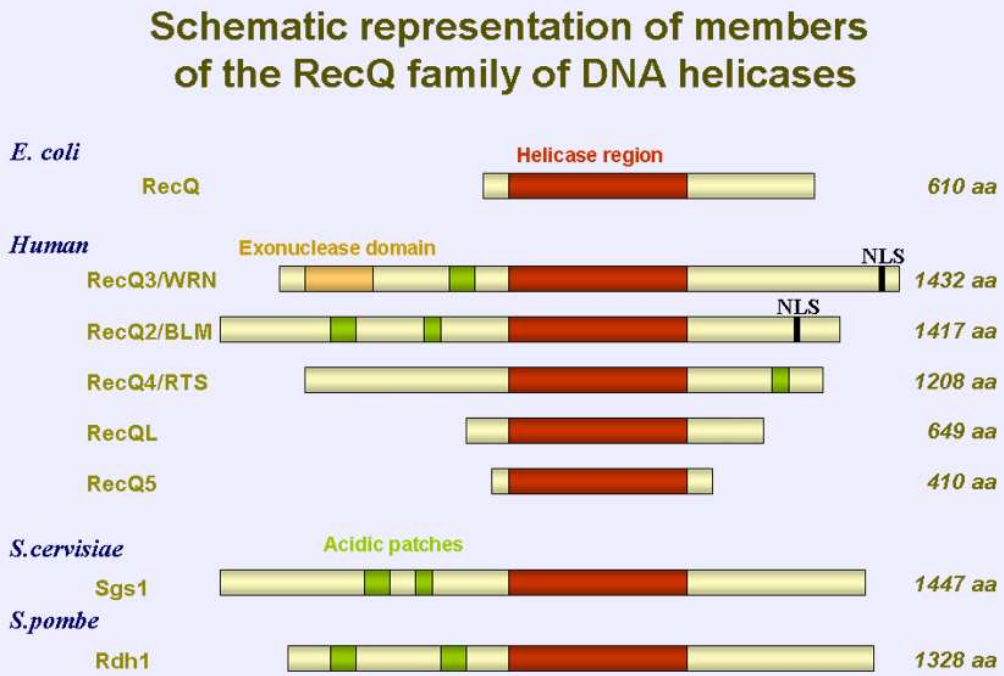
Description

Spans 6,46 kb; 21 exons; helicase domain is encoded by exons from 8 to 14.

Transcription

3,62 kb mRNA.





Protein

Description

1208 aa; 13,3 kDa; belongs to the RecQ subfamily of helicases and contains from aa 476 to 824 an helicase domain with a potential ATP binding site from aa 502 to 509, and the DEAH box from aa 605 to 608.

Expression

The RecQ4 gene is predominantly expressed in thymus and testis and at low levels in other organs such as heart, brain, placenta, pancreas, small intestine, and colon, indicating that the expression of RecQ4 gene is somewhat tissue-specific. The overall expression profile resembles that of the BLM gene. Interestingly, the expression of RecQ4 gene is partially upregulated in the G1/S phase of cell cycle.

Localisation

Nuclear.

Function

Suppresses promiscuous genetic recombination and ensures accurate chromosome segregation.

Homology

WRN/RECQ3, BLM/RECQ2.

Mutations

Germinal

See diagram of loss-of-function mutations in Rothmund-Thomson Syndrome patients.

Implicated in

Rothmund-Thomson Syndrome

Disease

Autosomal recessive disorder associated with genomic instability, cancer predisposition and premature ageing.

References

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